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ABSTRACT

This final report describes activities of the 4-year federally-funded Minnesota DeafBlind Assistance Project in meeting the following objectives: (1) provide technical assistance throughout the state; (2) deliver training to improve transitions from school to adult life for youth with deaf-blindness; (3) develop and implement procedures to locate and track individuals with deaf-blindness, with intense focus on early intervention and intervention for infants and toddlers; (4) facilitate family involvement and expansion of the family support program, with a focus on student advocacy; (5) provide educational training in current issues for individualized program development and inclusion of children and youth with deaf-blindness; (6) expand interagency collaborative planning and development; (7) develop a linking system with other Deaf-Blind Projects; (8) develop and disseminate resources of information services; (9) develop and implement first responder training for emergency care procedures for individuals with deaf-blindness; and (10) build linkages with the inter-tribal council, Indian Health Services, and Indian Education Program, and to collaborate with states in the Great Plains Regional Alliance to improve identification and services to American Indian children and youth with deaf-blindness. A bibliography from the DeafBlind Resource Library and a management manual for parents are attached. (CR)

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Minnesota DeafBlind Technical Assistance Project
Final Report: October 1, 1995 to September 30, 2000
(Includes 12 month no-cost extension)
Project Award #H025A50011

Project Director: George Holt

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During this grant cycle, Minnesota's children who are DeafBlind, their families and professionals working with them experienced great benefits from the Project. Families have received training to help them participate in their child's education as the leading member. They have been networking with one other via the Internet in multiple ways, including e-mail and listserves. Professionals have not only connected, but have received general deaf-blind and child specific training. On the whole, the state of Minnesota is more knowledgeable about children who are DeafBlind, the census has increased and systems change continues to evolve as a result of the Project leadership.

The Project has been fortunate to have a strong, consistent and experienced staff. The strength of the MN DeafBlind Project lies with its' dedicated and knowledgeable team members. When Project Director, George Holt(1995-1997), decided to leave his position at the Department of Children, Families and Learning to become an elementary principal, Carolyn Elliot took over as Grant Director(1997-1999). Carolyn was the state coordinator of the Regional Low Incidence Project, and facilitated the Low Incidence Work group through DCFL

In the second year of the grant, Coordinator Eric Kloos took a leave of absence for a year to receive a M.Ed in DeafBlind education at Boston College. Three skilled individuals shared some of the coordinator responsibilities in his absence. Sally Prouty and Dr.Sandra Davenport, who both worked for the Project for several years, increased their time and responsibilities. Additionally, the services of Paula Knutson, a DeafBlind Specialist for St. Paul schools, were purchased for the year.

Upon his return, in the third year of the grant Eric Kloos would take the role of Co-Director with Carolyn Elliot. Sally Prouty, who previously shared her position with the non-profit organization DeafBlind Services Minnesota (formerly FIND, Inc.), would put 100% of her energies in the Project as the Family Program Coordinator. In the carry-over year of the Grant, Carolyn Elliot retired, leaving vacant her position as Supervisor of the Low Incidence Disabilities Unit at the Department of Children, Families and Learning. Eric Kloos applied and was offered the position. Eric remains Director of the Project and his ability to influence policy and advocate for systems change on a state level could have huge implications for children who are DeafBlind in Minnesota. His leadership and experience are great assets to the Project. Sally Prouty became Coordinator and Cathy Lyle, the Educational Consultant. Cathy has 22 years experience working in all levels of education as resource teacher, itinerant teacher, and self-contained teacher for students who are deaf or hard of hearing. The Project has contracted with Intermediate School District #916 for the release of her time with the DeafBlind Project. The Project is also fortunate to have the expertise of Dr. Sandra Davenport, Pediatric Geneticist and Developmental Pediatrician, on staff. Team membership consisting of a parent, educator and pediatric sensory geneticist has led to the ability to view each child in a holistic manner. Numerous parents and individuals on

educational teams have commented positively on the team expertise provided to DeafBlind children and families in Minnesota.

The Project has benefited from the working relationship with our current fiscal host, the Educational Cooperative Service Unit in this grant cycle. The facilities, networking opportunities, dissemination possibilities, and the indirect costs have improved over previous fiscal hosts.

On the state level, the working relationship through the Department of Children, Families and Learning has been very beneficial. The support and networking done throughout the Department has been a significant factor in the increased statewide exposure of the Project. The Project director is the point-of-contact for the department for all deaf-blind policy and related issues. Additionally, collaborative projects focused on guidelines for the interpretation of FAPE for students with deaf-blindness, graduation standards, assistive technology, policy, monitoring and compliance and implementation of the IDEA-97. Project representation on the Low Incidence Workgroup for the Department of Children, Families and Learning as well as with the Regional Low Incidence Facilitators has been overwhelmingly positive. Work has focused on building capacity through statewide low incidence education issues. These coordinated efforts have resulted in greater exposure throughout the state and multiple new referrals for technical assistance. In addition the Project has been able to keep a finger on the pulse of statewide regional and local needs as they change throughout the year.

Multi-state collaboration with other state 307.11 projects provided successes in many areas. The Great Plains Regional Alliance (including Minnesota, North and South Dakota, Nebraska, Wyoming and Montana) met regularly, shared and disseminated information related to identification of students living on Native American Reservations. The Project will continue to collaborate on Model Demonstration Grants with the states of Utah and California. Project TRUST (TRansitions are centered around Understanding, with Support through Training), SPARKLE (Supporting Parent Access to Resources, Knowledge, Linkages and Education) and PRIIDE (Providing Resources through Interactive Instruction in DeafBlind Education) are grants through the SKI-HI Institute at Utah State University. Project SALUTE, another Model Demonstration Project awarded to California State University at Northridge, has included the MN Deaf-Blind Project to participate on their advisory and development committees by reviewing and field testing materials created to provide tactile learning opportunities for young children who are deaf-blind.

The Summer Transition Program has had a huge impact on the state and transition-aged students with deaf-blindness. The Project collaborated with State Services for the Blind and school districts to make this three-week training opportunity available for transition aged students who are deaf-blind. Six students successfully participated in years three and four of the grant. The youth participated in training, work experience and social opportunities while living independently in supervised dormitories. In the carry-over year of the grant four teens attended NTAC's Self-Determination Seminar in affiliation

with the American Association of the Deaf-Blind bi-annual conference in Columbus Ohio. Two additional MN teens participated in the AADB conference.

Minnesota has been instrumental in developing a unified look at Interveners for students who are deaf-blind. A "briefing paper" was developed with the collaboration of Linda Alsop of Utah, Robbie Blaha of Texas and Eric Kloos of Minnesota. NTAC plans to publish and disseminate this paper. In addition, with the support of NTAC and the Hilton/Perkins Foundation an advisory board was established to look into developing an Intervener training program in Minnesota. The advisory board met in year 3 and 4. As a result, in year 4, the first class on deaf-blindness, the Anatomy of the Eye and Ear videoconference course was taught in Minnesota and viewed live in Utah using distance technology.

This list of objectives and activities from the original grant application has been included for reference.

Objective 1: Provide technical assistance throughout the state.

Activity 1.1: To provide on-site consultation to educators, service providers, and families of children and youth with deaf-blindness.

Activity 1.2: To continue to conduct in-service training sessions for school personnel on a regional level.

Activity 1.3: To continue to develop and expand the Minnesota Deaf-Blind Resource library.

Activity 1:4: To create and maintain a data bank of individuals who have special knowledge and/or skills in working with individuals with deaf-blindness.

Activity 1:5: To develop and maintain a network of mentor teachers that work with children and youth with deaf-blindness.

The amount of on-site technical assistance in both homes and classrooms has increased six-fold in the past 4 years (from six in year 1 to forty-nine in year 4). The Project responds to requests by parents and/or educators for technical assistance. Efforts have been made to tailor presentations to the population they are presented to (for example, focusing on a certain etiology of deaf-blindness where appropriate, on child specific issues or topics such as communication methods or calendar systems.) This effort has been well received and gives guidance to each population (Activity 1.1). In collaboration with the Regional Low Incidence Facilitators, transdisciplinary teams from each of the 11 regions of the state have been identified and meetings held to discuss training needs. The goal is to build local capacity by supporting multiple teachers and professionals in each region of the state (Activity 1.2). The previous efforts to mentor teachers and identify knowledgeable professionals left the state with pockets of expertise, but also left large

areas of the state without local professionals to consult. The new design should provide more effective local capacity and mentors (See objective 9). Members of each regional transdisciplinary team receive training, are added to a data base and used as a resource to mentor more teachers (Activity 1.4 and 1.5). The Minnesota Deaf-Blind Resource library is continually updated with materials per teacher request and newly published resources (Activity 1.3).

Objective 2: Delivery of training to improve transitions from school to adult life in the community for youth with deaf-blindness.

Activity 2.1: To improve state-level planning and policy development for transitions of youth with deaf-blindness.

Activity 2.2: To enhance the participation of youth with deaf-blindness and their families in achieving successful transitions from school to community living.

Activity 2.3: To provide professional development and training that will help prepare youth with deaf-blindness for transition.

Activity 2.4: To collaborate with state level transition projects, such as the Minnesota Transition Project and Project Invest to support with cross-training in issues of deaf-blindness.

Activity 2.5: To support transition programs for high school students in the areas of vision and hearing to make them accessible to students with deaf-blindness. Two current programs include the State Services for the Blind Summer Transition Program and the Minnesota Resource Center for the Deaf and Hard of Hearing Post-Secondary Training.

Statewide representation through the State Transition Interagency Committee and the local Community Transition Interagency Committees was maintained this grant cycle (Activity 2.1, 2.2, 2.3, 2.4). In year two, planning and development of a summer option for transition-aged students with deaf-blindness was initiated. Heavy collaboration with local school districts, State Services for the Blind, DeafBlind Services Minnesota (a non-profit) and the Project produced excellent results. A pilot program was developed in year three to involve six students who are deaf-blind in the three-week Summer Transition Program (STP) for blind/visually impaired. Five students with deafblindness attended STP in year four. These students received deaf-blind specific training from leaders in the field on personal futures planning, recreation & leisure, and orientation & mobility (Activity 2.1, 2.2, 2.3). In addition to supporting the design and support for the deaf-blind portion of the program, the secondary goal was to provide deaf-blind specific training to staff and educators involved in the program (Activity 2.3, 2.4 & 2.5). Collaboration with Brian Abery of the University of Minnesota's Self-Determination Project the past three years included project design, assisting with focus groups, material review and access to families. Efforts to support NTAC's Self-Determination Summer

Seminar in the carry-over year resulted in Brian Abery presenting the self-determination curriculum to the students at AADB in Ohio. Seven teens with deafblindness from Minnesota attended the American Association for the Deaf-Blind national conference with four participating in the Self-Determination Seminar. The Project works closely with State Services for the Blind's two transition counselors and efforts are ongoing to coordinate and collaborate with various state transition initiatives (Activity 2.4 and 2.5).

Objective 3: Develop and implement procedures to locate and track individuals with deaf-blindness, with intense focus on early intervention and intervention for infants and toddlers who are deaf-blind.

Activity 3.1 To develop and implement procedures that will identify all individuals with deaf-blindness in the state.

Activity 3.2: To implement a systematic process of accurately collecting and sharing census information among state agencies.

Activity 3.3: To collaborate with the Vision Screen Project to develop and implement training to improve the assessment and screening services statewide.

The current census reports 208 children and youth with deaf-blindness in the state of Minnesota. There have been significant progress made in the area of early identification with a large increase in 0 – 2 year identification (Activity 3.1, 3.2). We are involved in deaf-blind awareness and training for the universal hearing screening that is voluntarily done in hospitals across Minnesota (Activity 3.1, 3.2). Census referrals are made from several sources including schools, State Services for the Blind, PACER Center, and DeafBlind Services Minnesota (Activity 3.2). We are working with the MN State Academy for the Blind and MN State Academy for the Deaf in the development of screening protocols (Activity 3.1, 3.2, 3.3). Collaboration with Dr. Sandra Davenport in the area of screening and identification resulted in new product development for training to improve the assessment and screening services statewide (Activity 3.1, 3.2, 3.3).

Objective 4: Facilitation of family involvement and expansion of the family support program, with a focus on student advocacy.

Activity 4.1: To continue to hold annual family enrichment weekend, called "Children Linking Families".

Activity 4.2: To continue to hold at least two educational and networking workshops for families yearly called "Children Linking Families".

Activity 4.3: To continue to empower families and expand the parents role in the Family Support Project so that they assume major administration of Children Linking Families events by end of FY 1996.

Activity 4.4: To develop and implement a statewide Family Mentor Program as part of the Family Support Project, to enhance the support network of families of children and youth with deaf-blindness.

Activity 4.5: To support families and consumers to attend workshops and conferences to foster leadership and empower with information of current best practices.

Activity 4.6: To continue to have representation and participation from parents and consumers on state teams and the Advisory Council.

Through the efforts of Sally Prouty, the Family Program Coordinator and now Project Coordinator, great strides have been made with families. She serves as a barometer to measure parent needs and satisfaction. A variety of activities have been conceived to encourage additional parent participation including a fathers fishing day, a monthly support group, a bi-weekly sign language/communication class in the evening and the Family Internet Connection (Activity 4.2, 4.3, 4.4, 4.5).

Families continue to find the annual "Children Linking Families" enrichment weekend a highlight of their year. Fifteen families attended the enrichment weekend the first year of this grant cycle increasing annually to a high of 23 families attending the last year of the cycle. Evaluations from the enrichment weekends are very positive with outcomes of 100% saying the weekend was valuable and their children were very happy and want to return annually. One parent summed it up on the evaluation saying, "The work you are doing is so appreciated not only for the weekend but for the year and also for the years to come. My prayer is that I will always be able to give back to the community like you have been for me and our family. You will never know how much direction and encouragement you have given" (Activity 4.1).

For the past three years, up to 26 families from throughout the state have been provided Internet access through the Family Internet Connection. The goal is to network families throughout the state and country to reduce isolation and provide connections to community and information as well as an excellent follow up to the family enrichment weekends. The technology allows parents to mentor one another from the comfort of their home and fits with their busy schedules. A six-page WWW resource list has been developed and widely distributed to families and professionals across the country. Additionally, work with SKI-HI on Project SPARKLE has increased the knowledge base of phase one families using e-mail and internet technology (Activity 4.4).

Numerous families have attended a variety of conferences this grant cycle including a National Parent to Parent Conference, National CHARGE conferences in Boston in 1997 and Texas in 1999, the New York/New Jersey Usher Syndrome conferences in 1997 and 1999, the Hilton/Perkins National Parent Conferences every year, and Closing the Gap Technology Conference. The Project sponsored an Usher Syndrome Family Fun Night in the carryover year. Two Midwest CHARGE workshops for families and professionals

were held with Minnesotans as well as residents from surrounding states in attendance. Parents brought back information to share through presentations, e-mail and newsletter (Activity 4.5). Parents are members of nearly all the regions' "First Responder Teams" or "Regional Network Teams" and are involved in the planning and implementation of a variety of Project events (Activity 4.6). Five parents are members of the Project Advisory Council (Activity 4.6) and we are fortunate to have a Minnesota parent represent the Midwest region for the National Family Association of Deaf-Blind(NFADB). A team of transition-aged teens and their interveners attended the summer Seminar on Self-Determination at the American Association for Deaf-Blind in Ohio in June 2000. Project staff will continue the Self-Determination training on the campus of a metropolitan community college where 8 deaf-blind teens(still on IEPs) are in attendance (Activity 4.2, 4.3).

Objective 5: Provide educational training in current issues for individualized program development and inclusion of children and youth with deaf-blindness.

Activity 5.1: To continue sponsoring the Deaf-Blind Training Institute, including scholarships for members of the Teacher Network.

Activity 5.2: To establish a statewide bulletin board for information on sensory disabilities on the electronic network, Specialnet, to provide direct information on deaf-blindness throughout Minnesota, to directors of special education, low incidence programs and teachers.

Activity 5.3: To collaborate with FIND, Inc. to develop and implement a school "intervener" model to support children with deaf-blindness in their school of choice.

Summer Institutes were offered the first three summers of this grant cycle focusing on early childhood in 1996, assessment and communication in 1997 and SKI-HI INSITE training in 1998. Two summer institutes were held in Greater Minnesota to reach teachers in rural areas of the state (Activity 5.1). Due to the changing identified training needs of professionals, summer training in 1998 and 1999 was held in partnership with the Summer Transition Program for Teens who are deaf-blind. Numerous deaf-blind experts were brought in to present to both the teens and professional staff (Activity 5.1).

The state of Minnesota dropped GTE as their carrier and listserve in education in 1998. As a response, and also in connection with the family Internet resources, the Project has worked to develop a new Internet home page. It will be further developed as a way to disseminate information through the Internet (Activity 5.2).

Work has begun in developing Intervener training material and a training program. With the successful completion of Anatomy of the Eye and Ear videoconference course with Interveners, teachers and parents in Minnesota and the SKI-HI Institute in 1999, continued collaboration with key players at Boston College and Hilton/Perkins is

ongoing. A monthly Intervener support group was initiated and offered through the Project during the 1999-2000 school year. There have been several presentations on the Intervener topic by project staff within this grant cycle including the Hilton/Perkins National Conference on Deafblindness (1997), National CHARGE conference (1997), the Project Directors meetings (1998 & 1999) and multiple schools and professionals networks within the state of Minnesota. A briefing paper has also been developed in collaboration with Utah, Texas, and Minnesota and will be distributed by NTAC (Activity 5.3). (attached)

Objective 6: Expand interagency collaborative planning and development.

Activity 6.1: To support interagency planning in the areas of identification and programming for Usher Syndrome and CHARGE syndrome.

Activity 6.2: To collaborate with other state projects in the areas of inclusion, transition and early childhood.

Activity 6.3: To continue to have interagency representation on the Advisory Council and have it serve as a lead group for information sharing on deaf-blind issues in the State of Minnesota.

Although the Vision Screen Project ended four years ago, the Project continues to work and has expanded the role of Dr. Davenport. Co-development of new resources in the areas of CHARGE Syndrome and Usher Syndrome continued throughout the grant cycle. This includes work on the "CHARGE Syndrome: A Management Manual for Parents" that was completed in 1999 (attached). Project staff were involved in the planning and coordination of the International CHARGE Syndrome Conference in July 1997. Dr. Davenport has also served as a regular consultant to the project for onsite technical assistance, providing a crucial bridge between the education and medical worlds. Project staff and families have attended the New York/New Jersey Usher Syndrome Family Conference in years 1997 & 1999 (Activity 6.1).

The Project director is involved in numerous policy committees in education, health, human services, social services and higher education on the state and national level. Project staff are members of the following advisory committees: State Services for the Blind Children Advocacy Board, DeafBlind Services Minnesota Adult Residential Services and the Children Youth & Family Services committees, the Southern Minnesota Psychological Assessment Services Advisory committee, Education sub-committee of the Minnesota Commission for Deaf/Hard of Hearing, and the Universal Newborn Screening Committee (Activity 6.2).

Interagency representation on the Advisory Council is strong. The Advisory Council continues to be the lead group for information sharing on DeafBlind issues in the state. Twenty-eight members represent a broad range including five parents, two consumers, education, rehabilitation, social services, health, and higher education. In addition,

members represent the National Family Association for Deaf-Blind, PACER Center, DeafBlind Services Minnesota (a non-profit) and the Institute on Community Integration at the University of Minnesota (Activity 6.3).

Objective 7: Develop a linking system with other Deaf-Blind Projects, nationally and internationally, to collaborate on development of project objectives.

Activity 7.1: To continue to work with TRACES to obtain technical assistance in the areas of functional assessment, self-advocacy, cultural issues, early intervention, and transition.

Activity 7.2: To disseminate project information to Deaf-Blind projects, nationally and internationally.

Activity 7.3: To attend national project directors meetings.

Activity 7.4: To participate in staff development opportunities, which will enhance the implementation of Project goals.

As follow up to the site review process (completed in 1996), staff have attended annual NTAC workshops since February 1997 including "Family Specialists - The Critical Link" (1997), "Parents Working with Parents - an Experiential Training" (1998), "Important Practices in Communication: Making it Happen at Home and in the Community" and "Distance Technology" (1999). NTAC sponsored SKI-HI INSITE training (1998) and co-sponsored the Self-Determination Seminar the summer of 2000 that four Minnesota teens attended. In year three of the grant, focus groups were organized to study the training needs for Interveners. Following the focus groups, national experts joined Project staff and Minnesota stakeholders in developing an advisory council to study the feasibility of establishing an intervener training program at the College of St. Catherine (Activity 7.1).

Collaboration and dissemination and has occurred with multiple projects around the country. Minnesota will continue to collaborate as a state partner with the SKI-HI Institute at Utah State University on model demonstration grants. Project TRUST focused on transition within early childhood. Project SPARKLE and PRIDE focus on using distance methods and CD-ROM technology to train families, teachers and interveners. Project SALUTE, another model demonstration project awarded to California State University at Northridge, includes the MN Deaf-Blind Project on their advisory and development committees involved in reviewing and field testing materials created to provide tactile learning opportunities for young children who are deaf-blind (Activity 7.2).

Project staff have presented information locally, nationally and internationally. Local conference presentations include; Deaf/Hard of Hearing Annual Teacher conference, Blind/Visually Impaired Annual Teacher conference, Early Childhood Coordinators

psychologists, assistive technology and the Council for Exceptional Children. National and International conference presentations include: 1997 National Conference on DeafBlindness, 1997 & 1999 CHARGE Syndrome Conferences, 1998 & 1999, Project Directors Meeting, 1999 International Conference on DeafBlindness in Portugal, 1999 Anatomy of the Ear and Eye: Etiologies of DeafBlindness videoconference, 2000 Great Plains Regional Alliance summer training, and NTAC's 2000 Strategies of Effective Family Technical Assistance training (Activity 7.2 & 7.4).

Project staff annually attend the Project Directors meeting (Activity 7.3) and have attended a multitude of local and national training opportunities including those listed above as well as two American Association for the Deaf-Blind bi-annual conferences and a Lilli Nielson workshop. Locally, training opportunities have included cane use with Joe Cutter, orientation and mobility with Joe Coiffi, recreation and leisure activities with Lauren Lieberman, personal futures planning with Janet Stevely, psychoeducational assessment via video conference with Harvey Mar and teaching visually impaired students in the new millennium with Dr. Egbert. Two project staff attended Boston College, one for a year while on leave of absence receiving a M.Ed. in DeafBlindess and the other for the three-week summer training program sponsored by Boston College and Hilton/Perkins (Activity 7.4).

Objective 8: Develop and disseminate resources of information services.

Activity 8.1: To disseminate the Advisory Council's guide to program development.

Activity 8.2: To make communication with the Project Accessible in multiple ways, through voice and TTY telephone, a toll-free 800 number, and the Internet and Specialnet networks.

Activity 8.3: To publish the deaf-blind newsletter, *The Informative Link* twice per year.

Activity 8.4: To continue the dissemination of Project brochures to teachers, special education directors, and low-incidence coordinators.

Activity 8.5: To continue to update and disseminate a resource library materials list to educators, families, and service providers.

The Advisory Council's guide to program development continues to be disseminated (Activity 8.1). The Project is accessible in multiple ways to better meet the needs of all who use our services. Toll-free telephone, voice and TTY, e-mail and Internet access all have increased the ways that people can communicate with the Project. The Internet has provided access to multiple national and international lists, including deaf-blind, blind, retinitis pigmentosa and others too numerous to mention. A World Wide Web Resource Guide has been developed and distributed locally and nationally (Activity 8.2). The

Guide has been developed and distributed locally and nationally (Activity 8.2). The *Informative Link* newsletter underwent a transformation and became the *DeafBlind Inquirer*(attached), in collaboration with DeafBlind Services Minnesota (DBSM). The last year of the grant cycle, our collaborative partner on the newsletter, DBSM, chose to publish their own newsletter to market their services as well as solicit financial support. (Project staff continue to write a regular column in their parent newsletter.) In year four, the Project collaborated with the University of Minnesota, Institute on Community Integration, to publish the *Impact* newsletter (attached), which focused on Deaf-Blindness and transition. Discussions are ongoing to decide the best avenue to provide information to education professionals, including review of possibilities for distribution using electronic formats (Activity 8.3). Dissemination of Project brochures (attached) and resources occurs at meetings, conferences, presentations, technical assistance visits and every opportunity possible (Activity 8.4). The Project is currently taking an inventory of books, videos, curriculum, journals, videotapes and CD-ROMs in the resource library. The Project plans to keep the library current, replacing old materials, updating resources and keeping duplicate copies of popular materials (Activity 8.5).

Objective 9: Develop and implement first responder training for emergency care procedures for individuals with deaf-blindness.

Activity 9.1: To implement the training of trainers for the first responder program.

Activity 9.2: To provide families of individuals with deaf-blindness with alternative emergency identification procedures.

This is the largest area of change, in terms of perceived need when the grant was written versus current need. The First Responder concept has shifted to focus on regional transdisciplinary teams of professional and strengthening that local capacity. Feedback from regional low incidence facilitators and the advisory council, supported by needs assessment, determined that the previous First Responder concept, and specifically the communication board, lacked clarity and utility for the original purpose. The redesigned First Responder concept is an attempt to get to the same end point, through a different system. Work with families will focus on the area of identification and communication in the community for safe travel, and the development of resources. The first responder program will provide a long-term group of professionals that will access many years of in-depth training to build Deaf-Blind expertise and capacity within each region of the state (Activity 9.1 & 9.2).

Objective 10: Build linkages with the inter-tribal council, Indian Health Services, and Indian Education Program, and to collaborate with states in the Great Plains Regional Alliance to improve identification and services to American Indian children and youth with deaf-blindness.

Activity 10.1: To identify tribal administration systems, including principle chiefs and educational leaders on each of the four BIA reservations in Minnesota that have independent school programs.

Activity 10.2: To promote collaboration with the BIA entities listed above to enhance dissemination of program information, identification information and assessment availability, and to determine direct service needs for all children and youth ages birth through 21 years with deaf-blindness.

Activity 10.3: To form a collaborative alliance with the states of the Great Plains (Montana, Wyoming, Nebraska, North Dakota, and South Dakota) to focus on communication, shared training and expertise to support children identified as deaf-blind who reside on American Indian reservation land.

Activity 10.4: To collaborate with tribal leaders and service providers to increase by 80% the number of children and youth identified as deaf-blind who live on American Indian reservation land.

Activity 10.5: To create a system of coordinated local, tribal, and governmental services to meet the needs of children and youth identified as deaf-blind residing on American Indian reservation land.

Contact was made in the area of screening and identification. Lynn Eidahl, a consultant to the Project, developed contacts with the Indian Health Services and the medical community (Activity 10.1 & 10.5). Brochures were developed and disseminated and a public service announcement was developed. Information about the Project and services has been forwarded to all schools serving American Indian children. To further contact and impact on reservation lands, the Project continues to explore and develop contacts for dissemination of information and representation at a local level (Activity 10.2). Members of the Great Plains Regional Alliance (including Minnesota, North and South Dakota, Nebraska, Wyoming and Montana) met at least annually to collaborate and share information with the outcome to collaborate on summer 2000 training in Montana for member states of the Alliance (Activity 10.3). In the area of identification, new questions specific to Native Americans were added to the census forms. In addition, the first native american student with deaf-blindness was identified in Minnesota in 1999 (Activity 10.4). Project staff traveled to the school, observed and provided on-site training for the staff. Lynn Eidahl, who had been the lead consultant, left the Project in 1998 to accept a position with the Department of Health in Early Childhood tracking, but the connections and capacity of the health service system remain.

A Discussion of Problems encountered and how they were solved.

There was a slight disruption with staff turnover. The director and coordinator positions, although stable when the grant started the new cycle, changed soon after and marked the first of several changes throughout the grant. George Holt, Director, left his position with

the Department of Children, Families and Learning to become principal of a local elementary school. With that, Carolyn Elliot, the state supervisor of low incidence projects assumed the responsibilities of co-director with Eric Kloos. Eric took a leave of absence for a year to receive a M.Ed. in Deaf-blind education at Boston College. While away, existing Project staff increased their time and added the services of Paula Knutson, a deaf-blind specialist for St. Paul Schools, whose time was purchased for the year. When Ms. Elliot retired, Eric applied and was offered her position with the Department of Children, Families and Learning. Eric remains the Director of the Project. Cathy Lyle, a teacher of the deaf and hard of hearing, was added to the staff as educational consultant. Currently, the Project and staff have a much greater capacity than at the beginning of the grant cycle. Staff expertise is the strength of the Minnesota DeafBlind Project.

ATTACHMENTS

Minnesota Developmental Timeline

Sandra L.H. Davenport, M.D.
Eric Kloos, M.Ed.
Sally Prouty

PURPOSE:

Most developmental tables and charts show developmental milestones without taking into account any sensory losses, hospitalizations, illnesses, educational interventions or major family events. The Minnesota Developmental Timeline shows the relationship of all these factors over time. The purpose is to understand more clearly the unique circumstances which affect a child's learning. This chart should be kept by the family with copies becoming part of a child's permanent educational and/or medical records. It should also be updated periodically on the parent's original with the chart copies being replaced as necessary. Some parents may decide they do not want to keep the original.

The chart is both succinct and approximate, i.e. exact dates and lengths of hospitalization, great detail on milestones are not necessary. The focus should be on the overall picture and the relationship of events. Details can be written on the back of the page or on separate sheets.

WHO SHOULD PROVIDE THE DATA:

Parents or other caretakers usually can provide the most of the information. Medical and educational professionals can help make the descriptions more accurate and precise and help fill in the informational gaps.

HOW TO CONSTRUCT THE CHART:

1. While a form is often provided and you can certainly use a handwritten form, you may also wish to develop your own chart using a drawing program such as Corel Draw or by using as a spreadsheet like Excel. The drawing programs produce pretty results but are more time-consuming to construct.The spreadsheet is more readily available and will make it easy to update the information yourself while keeping the information easy to read. Two drawbacks are that the age/date columns become uneven and the hospitalization lines will have to be drawn in by hand.
2. Enter child's name, birthdate and the date the chart was filled in at the top of each page used. Decide whether you want the hatch marks across the top of the chart to represent days, weeks, months or years. Sometimes having a whole year on one page will not give enough space to fill in all the important events. A young child might need one page to represent 6 months while an older child might need one page to depict the first 3 years of life. Write in the age at the larger marks and also put the date plus age at least twice on each page, e.g. 6 mo. (8/96). Having both makes it easier to fill in information. Sometimes parents remember an event occurred just after New Year's but would have to figure out how old the child was at that time.
3. Mark in all hospitalizations (see Figure 1). These are bars which span the entire page and are shaded or colored in. The reason for doing this is that children typically show delay or even regression in development during and sometimes after a hospitalization depending on the severity of the illness and the degree of perceived physical or

emotional trauma experienced. Hospitalizations therefore have a major impact on all areas of development.

4. Write in precise terms the reason for hospitalization or major illnesses (see Figure 2). For instance, PE tubes, Nissen, pneumonia, EUA (exam under anesthesia), etc. Explain procedures or abbreviations on the reverse side. Under Other Illness write in medical problems like ear infection, swallowing problem, etc. A line can be drawn from the description across the page to the date or age at which the problem resolved. For instance, swallowing problems might last from birth to 3 ½ yr. so the words are written at the birth line and a line with an arrow is drawn across the entire first page and subsequent page(s) ending at 3 ½ as shown in Figure 2.
5. Fill in developmental milestones (see Figure 3). Motor and Mobility includes both gross motor milestones and O&M (orientation and mobility) milestones if applicable. Under Vision, Hearing, Taste and Smell note what sensory stimuli the child responded to at different ages and give numbers for visual acuity, degree of hearing loss, etc. when tests were given. Also note when glasses, hearing aids, FM system at home or school, or any other devise was introduced. Use a line across the page to note how long the child used the device if it was discontinued. Use an asterisk or number to indicate an explanation on the reverse side as to how long the child used the device. For instance, a hearing aid might have been fitted at age 3 yr. but was only used during preschool Mon, Wed, Fri, for 2 ½ hr during the school year. A dotted or dashed line might indicate this kind of non-continuous use.
6. Under Educational Services note when teachers and therapists began and stopped working with a child. (See Figure 4) Use abbreviations like ECSE (early childhood special education), PT (physical therapy), etc. Note whether this is at home or school and duration of service, i.e. 2x/wk for 30 min. This kind of detail can be put in an explanatory note on the reverse side.
7. Major Family Events are entered next (see Figure 5). These should include anything that can affect development like the birth of another sibling, death of a grandmother, a move, foster placement, frequent changes in home health care aides, etc.
8. Color code major areas that need emphasis (see Figure 6). For instance, hospitalizations might be colored with yellow highlighter. Anything that affects hearing can be highlighted in pink such as hearing milestones, ear infections, PE tubes, deaf/hard of hearing teacher, deaf uncle moved into town. Please note that color coding does not show up on black and white copies so use this judiciously.

POSSIBLE USES FOR THIS CHART:

1. Summary for parents/guardians.
2. Summary of past history for child's education file. The chart can be amended periodically and available for all new staff to review.
3. Summary for medical file.
4. Summary for case managers in settings outside education or clinics.

Name _____ B.D. 3-8-94 Date: 10/5/98

Age (yrs) (mo.)	Birth	3-4-94	5/94	7/94	9/94	11/94	1/95	3/95	(1 yr)	5/95	7/95	9/95	11/95	1/96	3/96 (2 yr)
Ill or in Hospital	G-tube/Nissen Hrt cath-ASD VSD, PDA, hypo-plastic rt. vent. mild hydroceph			Heart surgery Hemi-Fontan		Respiratory infection-ICU on vent	Respiratory infection	Bowel obstruction	Scope airway	Lacerations x2			PE Tubes #1 Trach closure	Aspir. Barium Resp. arrest	
Other Medical	Swallowing problems	Oral stim. program started w/ unilat facial palsy	OT at home	Pala. limp 2due to GE reflux	Recurrent ear infections		On steroids stopped oral feed, "nasty" disposition		Started oral stim again						
Motor & Mobility								Rolled, did not stay on front				Scooted on back			
Fine Motor												Pick up lint Stack 2 blks			
Tactile				Markedly defensive to all touch. Started on going pro-gram of brushing											
Vision	Colobomas of both optic nerves LI>Rt			Tracked w/ both eyes when well							Reached for something on floor but would feel around for it			Glasses for farsighted ness helped w/ accuracy	
Smell							No definite reaction to any smell								
Taste							Wrinkles face to picante sauce & sour pickles								
Hearing		Turn to voice. Rt hearing aid used 1 hr/d but no in hospital								Add Ll hear-ing aid but hard to keep on floppy ears so use still inconsistent					
Communi-cation				Parents started learning sign lang				1st word = 'more' in sign							
Education/ Services	OT to home						SKI-HI lang teacher 1x/wk PT					ECSE x 6 mo. PT, OT, DHH continued at home during school year until preschool			
Family Events	No home nursing help										Moved to Minnesota				

Name _____ B.D. 3-8-94 Date: 10/5/98

Age (yrs) (mo.)	3/96 (2 yr)	5/96	7/96	9/96	11/96	1/97	3/97 (4 yr)	5/97	7/97	9/97	11/97	1/98	3/98 (5 yr)
Ill or in Hospital		Heart cath		Heart surg-ery: Fontan				Otoplasty Eye muscle surgery					
Other Medical	Swallowing problems; Recurrent ear infections									oral stim program at school			
Motor & Mobility		5-pt crawl Few steps holding on											Walked independently
Fine Motor				Held pencil fairly well w/ little drawing						Made circles w/ open bottoms			
Tactile										Refused to touch any-thing, e.g. fingerpaints			
Vision													
Smell	No definite reaction to any smell									Imitated smelling flowers but never discrim-inated among smells			
Taste													
Hearing									L hearing aid	FM unit at school			
Communi-cation	2-word sentences Increasing vocabulary & understanding		"Funny Daddy" on Father's Day										
Education/ Services									Summer school July	Started preschool w/ interpreter			
Family Events													

Name _____ B.D. 3-8-94 Date: 10/5/98

Age (yrs) (mo.)	3/98 (5 yr)	5/98	7/98	9/98	11/98	1/99	3/99 (6 yr)	5/99	7/99	9/99	11/99	1/00	3/00 (7 yr)
Ill or in Hospital	PE Tubes #2												
Other Medical	→	→	→	G-tube feeds but not aspirating									
Motor & Mobility		Falling - bruise on head		Can stand up in mid room Less falling									
Fine Motor			Makes triangle w/ round bottom	Letters-not interested. Draws face not body Closing circles									
Tactile				Now touches anything but she has to initiate touch									
Vision			Rt: 20/50 Lt: 20/60 Upper fields partly missing	Can ID each kind of flower by color									
Smell	No definite reaction to any smell →	→	→	→									
Taste													
Hearing		FM unit purchased for home use	Began responding more to voice										
Communi-cation			Using both sign and voice	Asking others to fingerspell everything. Uses 7-8 word sentences									
Education/ Services		Summer school July PT all summer for balance											
Family Events													

WWW Resource List

compiled for
Family Internet Connection families
by
the Minnesota DeafBlind Technical Assistance Project

If you know of a web site that may interest another family, please send the address to Sally Prouty at mndb@skypoint.com

HEARING LOSS

***Audiograms,** would you like to know how to read one? Try
http://www.earinfo.com

Hearing Loss: This web page provides links to a wide variety of sites dealing with hearing loss. Take a virtual tour of the ear, learn about cochlear implants, or get the latest on ASL. This large, comprehensive site can take you there.
http://www.gwha.com/projects/hear/link/linkbody.html

***Hearing Loss Information and Resources:** This very large web page was developed by a mother of a child with hearing loss. It contains a massive amount of information from hearing dogs to education with much in between.
http://members.tripod.com/listenup

(NIDCD) The National Institute on Deafness and Other Communication Disorders; Hereditary Hearing Impairment Resource Registry (HHIRR):
http://www.boystown.org/hhirr

VISION LOSS

Blind related information: http://www.hicom.net/%7Eoedipus/blind.html

BLIND-L is a newsgroup that provides support for people who are blind with a primary focus on equipment and Computers. To subscribe, send an e-mail to:

TO: **listserv@uafsysb.uark.edu**
SUBJECT: subscribe
BODY: subscribe blind-l (your first and last name) Then, just send it and follow the instructions for confirmation.

Retina International is a voluntary charitable umbrella association of 34 national societies each of which is formed by people with retinal degenerative diseases such as Retinitis Pigmentosa(RP), Usher Syndrome, Macular Degeneration and allied retinal dystrophies, their families and friends. **http://www.irpa.org/home.htm**

Retinal Degeneration Discussion List (RPLIST) This list is an internet forum for discssion, questions and opinions related to retinal degeneration. To subscribe to this list, send to:

TO: listserv@sjunvm.stjohns.edu
SUBJECT:

BODY: SUBSCRIBE RPLIST(your first name and last name)
or coontact the listowners for assistance: rplist@sjuvm.stjohns.edu

DEAFBLIND SPECIFIC

American Association of the DeafBlind: aadb@erols.com

***DB-LINK, The National Information Clearinghouse On Children Who are DeafBlind,** "identifies, coordinates, and disseminates information related to children and youth who are deafblind. Parents, service providers, administrators and others interested in services are invited to contact DB-LINK for information. DB-LINK is a collaborative effort of the American Association of the DeafBlind, Helen Keller National Center, Perkins School for the Blind, and Teaching Research."
http://www.tr.wosc.osshe.edu/dblink

***DeafBlind Children Home Page,** by Shelly Mackie, parent of a DeafBlind child. She says, "This page is being set up as support for parents of deafblind children. We face many challenges in our everyday lives looking after and communicating with our deafblind children. My goals for this page are to:

1. Make a place for parents to gain information
2. Make a place for parents to talk with other parents who face the same unique challenges.

It is a place to share our children's life stories so that we may learn from one another. On this page so far is the story of Cameron my son. There is also information about joining a listserv dealing specifically with the needs of deafblind children. I have included a chat line which can be accessed from the web site and used for parents to chat with one another about topics related to their deafblind children. I am very excited about his new project and hope that everyone will come and see our site and participate in making this the Deafblind Children's Homepage." **http://www.geocities.com/Heartland/Meadows/5939/**

DeafBlind Children ListServe, by Shelly Mackie, a parent of a DeafBlind child (see above) This listserv will be open to Parents, Intervenors, and Professionals working with Deafblind children. The object of this listserv is to provide support and encouragement to one another, as we care for these children and their many unique challenges. The way a listserv works is simple. You subscribe to the list, this allows you to receive e-mail from the list and send e-mail to the list. The listserver takes the e-mails and sends them to everyone else on the list. This list will take on an open discussion format, and will also have a question/answer format where people working in the field of deafblindness will be available to respond to specific questions. To join the listserv send an E-mail

TO: **requests@talklist.com**
SUBJECT: blank
BODY OF THE MESSAGE: Subscribe Children

DEAFBLND list (notice there is no "i" in the word DEAFBLND) is a listserve with people who share and support one another who are both visually and hearing impaired along with parents, siblings, spouses, friends and other professional who want to learn from the DEAFBLND list. To subscribe, write an e-mail to:

TO: **listserv@tr.wou.edu**
SUBJECT: subscribe
BODY OF THE MESSAGE: subscribe deafblnd (your first and last name)

*Note: To send a message to this list send to: **deafblnd@tr.wou.edu** or contact Randy Klumph at klumphr@wou.edu for assistance.*

Deaf-Blind Perspectives is a newsletter published three times a year by the Teaching Research division of Western Oregon University.
http://www.tr.wosc.osshe.edu/tr/dbp/index.htm

Helen Keller National Center, http://www.helenkeller.org

***National Family Association of the Deaf-Blind: www.NFADB.org**

***On-Line courses in DeafBlindness** are offered through the Great Lakes Area Regional Center for Deafblind Education. It is a federally funded project awarded to the University of Dayton that serves the states of Ohio and Wisconsin. Online courses are:

Understanding Deafblindness: Overview - DB501
Understanding Deafblindness: Communication - DB502
Understanding Deafblindness: Orientation and Mobility - DB503

DB501, DB502, and DB503 are parts of a three course series on deafblindness offered through SSCOnline. These three courses cover the topics of communication, orientation and mobility, and how they relate to learners with vision or hearing impairment or multiple/severe disabilities. **http://www.ssco.esu.k12.oh.us/deafblind.html**

PARENT ADVOCACY

Advocacy Resources on Children with Disabilities: The Judge David L. Bazelon Center for Mental Health Law is a nonprofit legal advocacy organization based in Washington D.C. Our name honors the federal appeals court judge whose landmark decisions pioneered the field of mental health law, and our advocacy is based on the principle that every individual is entitled to choice and dignity. For many people with mental disabilities, this means something as basic as having a decent place to live, supportive services and equality of opportunity. **bazelon.org/children.html**

***PACER Center (Parent Advocacy Coalition for Educational Rights)** is founded on the concept of Parents Helping Parents. This Minnesota site features newsletters, articles on disability issues, federal and state legislation and more.
http://www.pacer.org

Special Education Advocacy you'll find articles, cases, newsletters, and other essential information about special education law and advocacy. The Special Ed Advocate has grown into an essential base for parents, attorneys, educators, and advocates. Parents, educators, experts, and attorneys come here for up-to-date information about effective advocacy for children with disabilities.
http://www.wrightslaw.com/mainpage_library.htm

PARENT SUPPORT

DREAMMS for Kids, Inc. (Developmental Research for the Effective Advancement of Memory and Motor Skills) is a non-profit parent and professional service agency, specializing in Assistive Technology related research , development, and information dissemination. DREAMMS is committed to facilitating the use of computers, assistive technologies, and quality instructional technologies for students and youth with special needs in schools, homes and the community.
website: www.dreamms.org or email: info@dreamms.org

Exceptional Parent Magazine an on-line magazine for families of children with disabilities. **www.eparent.com**

***Family Village:** At this website you can not only get information on deafblindness specifically, but you can find other websites as well, subscribe to a listserve where parents (and some professionals) can talk to each other, and access a library with information about a variety of other syndromes and rare disorders. They even have a Library (lots of info), a House of Worship, a Sports and Recreation Area, a Community Center, and a Bookstore. **http://www.familyvillage.wisc.edu/**

Family Voices: "We are families from throughout the United States who have children with special health needs. We are also caregivers, professionals, and friends whose lives have been touched by these children and their families. We are a diverse group, representing a wide variety of children, health conditions, families, and communities. Our concern for children brought us together". **www.familyvoices.org/**

Katlyn's Hope provides educational scholarships for DeafBlind children across the country. The scholarships have been used for intervener consultation, in-home training, educational toys and materials, assistive devices such as hearing aids, glasses, canes or other educational necessities. **www.idir.net/~khope/**

MUMS, is a national Parent to Parent organization for parents or care providers of a child with any disability, disorder, chromosomal abnormality or health condition. MUMS mission is to provide support to parents in the form of a networking system that matches them with other parents whose children have the same or similar condition. **http://www.waisman.wisc.edu/~rowley/mums/home.htmix**

Our-Kids list has over 900 people world-wide and is not specific to any single disability. However, things like feeding problems, therapies, adapted toys, etc. are common to many children with differing diagnosis so there is a lot of help available in the sharing of ideas. To subscribe, send an e-mail to:

TO: **LISTSERV@MAELSTROM.STJOHNS.EDU**
SUBJECT: <include anything here>
BODY: subscribe our-kids

OR

TO: **LISTSERV@MAELSTROM.STJOHNS.EDU**
SUBJECT: <include anything here>
BODY: subscribe our-kids-digest

Special Child: This site was developed by Lisa Baker, a mother of a child with special needs named Allison, and her brother/associate Troy Weiss. To provide educational resources and emotional support to parents and care-givers of children, adolescents, and young adults with disabilities. **www.specialchild.com**

***Texas School for the Blind and Visually Impaired** has a very large web site that includes a Family Discussion Room. It provides a place for families of children with visual impairments or deafblindness to connect with each other and to share information, issues, ideas, and resources. Since many of you may want to connect with another family who has a visually impaired or deafblind child like your own. (click on "contents" to see what other parents have posted) The site also contains copies of their excellent newsletter called See-Hear Newsletter - A quarterly newsletter for families and professionals on visual impairments and deafblindness. **www.tsbvi.edu**

SPECIAL EDUCATION

NCPSE, National Clearinghouse for Professions in Special Education, "provides information on recruitment and retention and overall supply of professionals in special education and related professional fields, with a particular focus on individuals with disabilities and those from culturally/linguistically diverse communities. It maintains a listing of programs of study in colleges and universities at both undergraduate and graduate levels, and provides resources for financial aid, non-traditional training programs, alternative certification, and job banks, as well as providing specific education career information." **http://www.cec.sped.org/ncpse.htm**

NICHCY, National Information Center for Children and Youth with Disabilities, was established by Congress. It is "an information and referral center that provides information on disabilities and disability-related issues as well as referrals to a wide network of specialists from agencies and organizations across the county. The focus is on education and children and youth, ages birth to 22 years."
http://www.aed.org/nichcy

HEALTH CARE

***NORD, National Organization of Rare Disorders** is the only organization of its kind--a unique federation of more than 140 not-for-profit voluntary health organizations serving people with rare disorders and disabilities. Thousands of affected individuals and their families--as well as support groups, health care and human service professionals, and advocates for people with rare disorders and disabilities--rely on NORD's assistance and leadership. NORD is a charity and continues its mission through the kindness and generosity of its our donors. **http://www.rarediseases.org**

Sensory Integration Sensory Integration International website. They have an excellent guide called "A Parent's Guide to Understanding Sensory Integration". It's only 15 pages and $2.00 and MOST helpful. **http://home.earthlink.net/~sensoryint/**

Sensory Integration Products: http://www.southpawenterprises.com

SYNDROMES

***CHARGE Syndrome:** information on CHARGE Syndrome, email list, Internet links, pictures of CHARGE kids, and the newest edition, a CHARGE chat room. **http://www.geocities.com/Heartland/1220** The E-mail list for families is a great place for support, information, and contact with other CHARGE families. To subscribe to the list, send an e-mail to:

TO: **CHARGE-subscribe@one.list.com**
SUBJECT:
BODY: subscribe charge-l (your e-mail address) *go to member user center link on left side of page.* **or** contact Casey Fisher, listowner, at: **charge@saber.net**

CHARGE Syndrome Foundation: www.chargesyndrome.org provides information about CHARGE syndrome as well as links to other sites.

Cornelia de Lange Syndrome (CdLS)Foundation, Inc's website include diagnosis protocol, a publications list, information about workshops and activities, links and more. **http://cdlsoutreach.org/**

National Congenital CMV Registry offers information on congenital CMV and a parent-to-parent connection. **http://www.bcm.tmc.edu/pedi/infect/cmv/**

***Usher Syndrome:** **http://www.boystown.org/hhirr/usher.htm**

http://www.geocities.com/hotsprings/7815

http://www.eng.dmu.ac.uk/~hgs/deafblind

***Usher Syndrome List serve**, send an e-mail to:
TO: **USHERS-SUBSCRIBE@onelist.com**
SUBJECT: subscribe
BODY: subscribe usher-list (your email address)
If you have difficulty subscribing, contact the list-owners(Randall Pope and Mark Graffis) at Ushers-owner@onelist.com

MISCELLANEOUS

American Sign Language (ASL) practice web site:
commtechlab.msu.edu/sites/aslweb/browser.htm

Government, both federal and state web sites.
MN Senate: **http://www.senate.leg.state.mn.us/**
MN House: **http://www.house.leg.state.mn.us/**
US Congress: **http://thomas.loc.gov/**
US Senate: **http://www.senate.gov/**
US Senators Email addresses: **http://www.senate.gov/senator/membmail.html**

Growth Hormone information, **http:www.magicfoundation.org** and **www.psigroup.com/dg/10760e.htm**

Intervenor List serve, this listserv is open to parents, Intervenors, and professionals. It will take on an open discussion format, and will also have a question/answer format where people working in the field of deafblindness (particularly congenital and early acquired deafblindness) will be available to respond to specific questions. The DB Intervenor Listserv is up and running. To Subscribe: Send an e-mail to:
TO: **DB_Intervenor@listserv.sd38.bc.ca**
SUBJECT: Type "subscribe.
BODY: leave blanK

John Tracy Clinic have worldwide services including correspondance courses in communication, auditory learning, speech etc. .**http://www.johntracyclinic.org/**

Note: The sites with an asterisk(*) are the most recommended.
The underlining was used as a way to highlight, do not underline when using these addresses.

The Family Internet Connection of the Minnesota DeafBlind Project is a statewide project committed to decreasing the isolation of families who have a child with a combined vision and hearing loss by providing computers, modems and information. The Family Internet Connection is not responsible for content within these internet sites. Comments, corrections and information are welcome. Revised 5/99.

Bibliography

Deafblind Resource Library

Minnesota Deafblind
Technical Assistance Project

FOR INFORMATION:
MINNESOTA DEAFBLIND
TECHNICAL ASSISTANCE PROJECT
Metro ECSU
4001 Stinson Blvd. NE, Suite 210
Minneapolis, MN 55421
612-706-0801 x106, x117 or x116
Fax: 612/706-0801
1-800-848-4905
TTY 612-706-0808
Internet: mndb@skypoint.com

Welcome to the Resource Library of the Minnesota Deafblind Technical Assistance Project

The Deafblind Resource Library is designed to help educators, parents, and other community members obtain information for improving the quality of life of people with disabilities. We only ask that you use the check out procedures given and return resources in a timely manner so that others may use them.

ADDRESS: Minnesota Deafblind Technical Assistance Project
Metro ECSU
4001 Stinson Blvd. NE, Suite 210
Minneapolis, MN 55421

PHONE: 612/706-0801, x106, x116, x117
FAX: 612/706-0811
1/800/848-4905
TTY: 612/706-0808

INTERNET: mndb@skypoint.com

HOURS: 8:00 a.m. - 4:30 p.m., Monday through Friday

VISITING THE LIBRARY: The library is located at Metro ECSU on the second floor of the address given above. We welcome you to visit our library. Please ask for directions for checking out resources.

USING THE LIBRARY BY MAIL: If you cannot visit the library, most materials can be mailed on loan to you. When calling to check out an item, please give the name of the resource needed, your name, address, phone number, and school district. Resources are mailed Library rate, and the Project will cover the cost of postage when mailing items to you. Return postage is your responsibility. Please use a padded envelope or sturdy box when returning materials.

LOAN SCHEDULE: Videos and printed materials may be on loan for 4 weeks.

Bibliography

DeafBlind Resource Library

Minnesota Deaf-Blind Technical Assistance Project

About the library...

The DeafBlind Resource Library offers over 400 books, curricula, videotapes, and other materials. Though much of the library focuses on resources related specifically to individuals with deafblindness, it also includes many materials with broader application in the disabilities field. This educational library is designed to help educators, parents, and other community members obtain information to help improve the quality of life of people with deafblindness and other disabilities. We only ask that you check out the materials by following the directions posted and bring them back in a timely fashion so that others may utilize them.

Materials in the library have been assigned category codes that appear in **bold** in bibliography entries on the first indented line of the entry and on the library card in the pocket. Categories were assigned if the resource contained a significant amount of information on that topic. Categories are as follows:

Code	Category
A	Assessment
B/VI	Blind/Visually Impaired
C	Communication
DD	Developmental Disabilities
D/HH	Deaf/Hard of Hearing
EC	Early Childhood
F	Family Focus
H/M	Health/Medical
I	Integration/Inclusion
M	Mobility
S	Sensorimotor Skills
T	Transition to Adulthood

Access To Mass Transit for Blind and Visually Impaired Travelers. Uslan, Mark M., Peck, Alec F., Wiener, William R., and Stern, Arlene. New York, NY: American Foundation for the Blind. 1990.

B/VI M

The contents of this text includes mass transit for blind and visually impaired persons, issues in rapid rail and bus travel, and orientation and mobility training. (179 pp.)
2 copies.

Acquisition of Basic Sensory and Motor Skills within Natural, Integrated Contexts with Students Facing the Most Serious Challenges. Gee, Kathleen, Graham, Nan, Lee, Mellanie, and Goetz, Lori. San Francisco, CA: San Francisco State University.

S

(29 pp.)

Administrative Guidelines: Alemeda Unified School District Guidelines for a Community Intensive Model. Doering, Katherine, Hill, Rina, and Lee, Melanie.

DD I

From Community Intensive Programs for Students with Sensory Impairments Project.

Advancements: An Implementation Guide to a Community-Based Vocational Training Program for Deaf-Blind Youth. Goros, Denise L. and Kowalski-Glickman, Mary. Watertown, MA: Perkins School for the Blind. 1983.

I T

After School...Then What? The Transition to Adulthood. Lehr, Susan. Syracuse, NY: The Center on Human Policy, Syracuse University. 1986.

DD T

Prepared for the Technical Assistance for Parent Programs (TAPP) Project, Boston, MA.
(62 pp.)

American Deaf Culture: An Anthology. Wilcox, Sherman (Ed.) Burtonsville, MD: Linstok Press. 1989.

D/HH

American Indian Concepts Concerning Health and Unwellness by Carol Locust, Ph.D Native American Research and Training Center, University of Arizona Book

American Indian Cultural Perspectives on Disability by Jennie R. Joe, Ph.D. and Dorothy Miller, DSW, Native American Research and Training Center, University of Arizona

American Indians: Answers to Today's Questions. Utter, Jack. Lake Ann, MI: National Woodlands Publishing Company. 1993.

The Answer Book: Job Search Strategies for Students with Disabilities.

DD T

(135 pp.)

Are You Blind?: Promotion of the Development of Children Who Are Especially Developmentally Threatened. Nielsen, Lilli. Copenhagen, Denmark: SIKON. 1990.

B/VI

(109 pp.)

Art of the Eye: An Exhibition on Vision. Minneapolis, MN: FORECAST. 1986. (96 pp.)

Assessing Infants and Preschoolers with Handicaps. Bailey Jr., D.B. and Wolery, M. Merrill Publishing Company. 1989.

A C DD EC S

This book includes: assessment and its importance in early intervention; procedural considerations in assessing infants and preschoolers with handicaps; Child Find and screening issues; screening and assessing sensory functioning; assessment of behavior, cognition, motor skills, communication skills, self-care skills, social skills, and play skills; and using assessment information to plan instructional programs. (516 pp.)

Assessing the School-Age Student w/ Dual Sensory & Multiple Impairments (Ages 6-15), Assessment Guidelines, Volume 2, June E. Downing, Ph.D. 2 copies

Assessing the Transition Needs of Young Adults w/ Dual Sensory and Multiple Impairments, Assessment Guidelines, Volume 3, 2 copies

Assessing Young Children w/ Dual Sensory & Multiple Impairments (Ages birth to five) Assessment Guidelines Volume 1, Ellin Siegel-Causey, Ph.D., University of Nebraska-Lincoln, 2 copies

Assessment: Best Practices for Assessing Students With Impaired Hearing. Kyllo, Valerie Klansek, Enfiejian-Hoekstra, Valerie, McAnally, Patricia L., and Hoekstra, Al. Faribault, MN: Minnesota Resource Center, Hearing Impaired. 1988.

A D/HH

(70 pp.) 2 copies.

An Assessment Instrument for Families: Evaluating Employment for Individuals with Deaf-Blindness. Helen Keller National Center-Technical Assistance Center. 1995.

A T

(9 pp.)

Assessment of Individuals with Severe Handicaps: An Applied Behavior Appproach to Life Skills Assessment. Browder, Diane M. Baltimore, MD: Paul H. Brookes Publishing Co. 1986.

A DD

This book covers the assessment of critical functions and life skills in persons with severe handicaps. Using applied behavior analysis, it examines the assessment of motor skills, communication, academics, and social behavior in the context of developing these skills across life domains. (296 pp.)

Auditory Assessment and Programming for Severely Handicapped and Deaf-Blind Students. Bay Area Severely Handicapped Deaf-Blind Project.

A

Auditory Training in the Perkins Deaf-Blind Department. Robbins, Nan. Watertown, MA: Perkins School for the Blind. 1964.

Augmentative Communication for Children with Deaf-Blindness: Guidelines for Decision-Making. Cress, Cynthia J., Mathy-Laikko, Pamela, and Angelo, Jennifer. Monmouth, OR: Communication Skills Center, Oregon State System of Higher Education.

A C (36 pp.) 3 copies.

Basic Skills for Community Living/A Curriculum for Students with Visual Impairments and Multiple Disabilities/Texas School for the Blind and Visually Impaired.

Begin the Between: Planning for the Transition from High School to Adult Life. Matuszak, Trish, Langel, Patty, Goldberg, Marge, and Goldberg, Paula. Minneapolis, MN: PACER Center, Inc. 1992.
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(41 pp.)

Behavior Disorders of Children and Adolescents. Cullinan, Douglas, Epstein, Michael H., and Lloyd, John Wills. Englewood Cliffs, NJ: Prentice-Hall, Inc. 1983.
(338 pp.)

Behavior Management: An Alternative Approach. McCarthy, Michael.

Better IEPs. Bateman, Barbara D. Cresswell, OR: Otter Ink. 1992.
(135 pp.)

Body Image and the Severely Handicapped Rubella Child. Guldager, Virginia. Watertown, MA: Perkins School for the Blind. 1970.

Body Skills: A Motor Development Curriculum for Children. Werder, Judy K. and Bruininks, Robert H. Circle Pines, MN: American Guidance Service. 1988.
A S
Curriculum kit.

A Book About Me, Signed english book for children, 16 pages.

The Braille Monitor. (February, 1996). Baltimore, MD: National Federation for the Blind.
B/VI
Journal published by the National Federation for the Blind.

Breaking Barriers: How Children and Adults with Severe Handicaps Can Access the World Through Simple Technology. Levin, Jackie and Scherfenberg, Lynn. Minneapolis, MN: ABLENET. 1986.
(66 pp.)

Brothers, Sisters, and Special Needs: Information and Activities for Helping Young Siblings of Children with Chronic Illnesses and Developmental Disabilities. Lobato, D.L. Baltimore, MD: Paul H. Brookes Publishing Co. 1990.
DD EC F H/M
This text has an explicit curriculum and activity guide designed especially for children 3 to 8 years. It offers a series of workshops to help siblings get to know their own personal strengths, recognize positive characteristics of their handicapped brother or sister and other family members, and learn ways of coping constructively with stressful family events. (213 pages) 2 copies.

(Learners Companion Packet for) Brothers, Sisters, and Special Needs: Information and Activities for Helping Young Siblings of Children with Chronic Illnesses and Developmental Disabilities. Lobato, D.L. Baltimore, MD: Paul H. Brookes Publishing Co. 1990.

Brothers & Sisters: A Special Part of Exceptional Families. Powell, Thomas H. and Ogle, Peggy Ahrenhold. Baltimore, MD: Paul H. Brookes Publishing Co. 1985.

DD F

This book offers advice to parents and professionals on techniques and services that can help non-handicapped siblings better understand their unique feelings and circumstances. (226 pp.)

The Business of Living with Retinitis Pigmentosa "Dealing w/ the Threat of Loss" by Dorothy Stiefel Booklet No. 1 2 copies

The Business of Living with Retinitis Pigmentosa "Stress and Well-Being" by Dorothy Stiefel Booklet No. 1

Calendars/Reactive-Interactive Planning Systems for Developing Communication with Students with Deafblindness A Resource Manual for Professionals
Ann Gloyn, B.A., BPhEd., B.Ed.

Callier-Azusa Scale. Stillman, Robert (Ed.), et. al. Dallas, TX: South Central Regional Center for Services to Deaf-Blind Children and The University of Texas at Dallas, Callier Center for Communication Disorders. 1978.

A C DD EC S

This is a developmental scale specifically designed to aid in the assessment of deaf-blind children and children with severe and profound handicaps. It is especially comprehensive at lower developmental levels. (81 pp.)

The Case Management Team: Building Community Connections. Lippert, Toni. St. Paul, MN: Developmental Disabilities Program of the Metropolitan Council. 1987. (78 pp.)

Changing Inappropriate Sexual Behavior: A Community-Based Approach for Persons with Developmental Disabilities. Griffiths, Dorothy M., Quinsey, Vernon L., and Hingsburger, David. Baltimore, MD: Paul H. Brookes Publishing Co. 1989.

DD

This text covers treatment of inappropriate behavior displayed by persons with developmental disabilities. (209 pp.)

CHARGE SYNDROME: A Management Manual for Parents/ CHARGE Syndrome Foundation, 4th International Conference, Houston, Texas, July 23 - 25, 1999

(The Child Care Series) Book 1: Baby is Here! St. Paul, MN: HIPP, St. Paul-Ramsey Medical Center and MELD. 1986.

(The Child Care Series) Book 2: Feeding Your Child. St. Paul, MN: HIPP, St. Paul-Ramsey Medical Center and MELD. 1986.

(The Child Care Series) Book 3: Healthy Child/Sick Child. St. Paul, MN: HIPP, St. Paul-Ramsey Medical Center and MELD. 1986.

(The Child Care Series) Book 4: Safe Child and Emergencies. St. Paul. MN: HIPP, St. Paul-Ramsey Medical Center and MELD. 1986.

(The Child Care Series) Book 5: Baby Grows. St. Paul, MN: HIPP, St. Paul-Ramsey Medical Center and MELD. 1986.

(The Child Care Series) Book 6: Baby Plays. St. Paul, MN: HIPP, St. Paul-Ramsey Medical Center and MELD. 1986.

Choosing Options and Accommodations for Children: A Guide to Planning Inclusive Education. Giangreco, Michael F., Cloninger, Chigee J., and Iverson, Virginia Salce. Baltimore, MD: Paul H. Brookes Publishing Co. 1993.

A DD F I

This text describes the COACH program, a practical tool designed to operationalize inclusive educational practices for students with severe and profound disabilities. This program incorporates a strong sense of family priorities. (189 pp.)

Collaboration in the Schools: An Inservice and Preservice Curriculum for Teachers, Support Staff, and Administrators (Instructors Manual). West, J. Frederick, Idol, Lorna, and Cannon, Glenna. Austin, TX: PRO-ED. 1989.

(306 pp.)

Communication Assessment and Intervention for Adults with Mental Retardation. Calculator, Stephen N. and Bedrosian, Jan L. (Eds.) Boston, MA: Little, Brown and Co., Inc. 1988.

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This book gives an overview of mental retardation, characteristics of adults with mental retardation, program considerations, and communication assessment and intervention strategies. (359 pp.)

Communication Development in Young Children with Deaf-Blindness: Literature Review. Bullis, Michael and Fielding, Glen.(Eds.). Monmouth, OR: Communication Skills Center for Young Children with Deaf-Blindness, Oregon State System of Higher Education. 1988.

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(261 pp.) 3 copies.

Communication in Sign Language: A Series of Lessons for Beginning Signers. Clark, Yvonne L. and Clark, Thomas C. Logan, UT: Hope Inc. 1986.

C

(138 pp.) 2 copies.

Communication Intervention for Learners with Dual Sensory Impairments: A Team Approach. Stremel, Katherine and Wilson, Rebecca. MS: University of Southern Mississippi.

C

Communication Systems and Routines: A Decision Making Process. Stremel, Katherine, et. al. MS: University of Southern Mississippi. 1990.

C

Communication with Learners Who Are Deaf-Blind (conference manual). Perkins National Deaf-Blind Training Project and Texas Tech in collaboration with 307.11 State and Multi-state Deaf-Blind Services. 1995.

C

Manual from the conference held July 16-20, 1995. Presenters included Barbara McLetchie, Debbie Feeley, and Susan Bruce Marks.

Community-Based Curriculum: Instructional Strategies for Students with Severe Handicaps. Falvey, M.A. Baltimore, MD: Paul H. Brooks Publishing Co. 1986.

DD I

Outlines issues to be dealt with in designing and implementing community-based instruction for students with severe handicaps. It also describes preferred educational practices for teaching functional and age-appropriate skills in integrated settings, and provides practical recommendations for developing curricula individualized to match particular student needs. (241 pp.)

Community-Based Living Options for Young Adults with Deaf-Blindness: Philosophy, Directions, and Strategies. Barrett, Stephen S., Carr, Theresa, S., and Covert, Angela M. (Eds.). Sands Point, NY: Helen Keller National Center for Deaf-Blind Youth and Adults. 1987.

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(404 pp.) 3 copies.

Community Guide for Deaf-Blind and Severely Multi-Handicapped Students. Stoelting Company.

C DD I M

The guide has four components: Communication Skills, Techniques of Daily Living, Orientation and Mobility, and Sensory Stimulation.

Community Integration for People with Severe Disabilities. Taylor, Stephen J., Biklen, Doug, and Knoll, J. Teachers College Press. 1987.

DD I

This book provides a comprehensive review of issues involved in integrating people with severe handicaps into the community. (231 pp.)

Community Recreation and Persons with Disabilities: Strategies for Integration. Schleien, Stuart J. and Ray, M. Tipton. Baltimore, MD: Paul H. Brookes Publishing Co. 1988.

DD I

This text discusses community recreation and services for person with disabilities. (277 pp.) 2 copies.

A Companion Manual for Minnesota Rule 3525.2925: The Use of Behavioral Interventions with Pupils. St. Paul, MN: Minnesota Department of Education. 1992.

2 copies.

Competencies for Teachers of Learners Who Are Deafblind, Perkins National Deafblind Training Project, Barbara A.B. McLetchie & Marianne Riggio

Competitive Employment: New Horizons for Severely Disabled Individuals. Wehman, Paul. Baltimore, MD: Paul H. Brookes Publishing Co. 1981.

A DD T

The contents of this book include: assessment of employability; training for competitive employment; placement; job retention; employer, co-worker, and parent perceptions of workers with disabilities; development of nonvocational skills; analysis and evaluation of the failures in competitive employment; and model programs in competitive employment. (259 pp.)

Conference on Deafblindness

Living & Learning: A Lifelong Adventure, May 8-11, 1996

Count and Color, signed english book for children, 16 pages

Curriculum for Daily Living. Watertown, MA: Perkins School for the Blind. 1978.
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Curriculum Guide for Deaf-Blind and Severely Multi-Handicapped Students-Component I: Communication Skills. Chicago, IL: Stoelting Company. 1985.
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3 copies.

Curriculum Guide for Deaf-Blind and Severely Multi-Handicapped Students-Component II: Techniques of Daily Living. Chicago, IL: Stoelting Company. 1985.
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Curriculum Guide for Deaf-Blind and Severely Multi-Handicapped Students-Component III: Orientation and Mobility Chicago, IL: Stoelting Company. 1985.
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Curriculum Guide for Deaf-Blind and Severely Multi-Handicapped Students-Component IV: Sensory Stimulation Chicago, IL: Stoelting Company. 1985.
3 copies.

Curriculum Planning and Development for Deaf-Blind Severely Handicapped Students. Rocksund, Jill.
From the Career Exploration Training Project in collaboration with Woodhaven School, Inc. (34 pp.)

Deaf -Blind Children: A Program for Evaluating Their Multiple Handicaps. Curtis, Scott, Donlon, Edward T., and Wagner, Elizabeth (Eds.). New York, NY: American Foundation for the Blind. 1970.
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(172 pp.)

Deaf-Blind Education: Developing Individually Appropriate Communication and Language Environments, Book A. Robbins, Nan (Ed.). Watertown, MA: New England Regional Center for Services to Deaf-Blind Children. 1983.
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Deaf-Blind Education: Developing Individually Appropriate Communication and Language Environments, Book B. Robbins, Nan (Ed.). Watertown, MA: New England Regional Center for Services to Deaf-Blind Children. 1983.
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Deaf-Blind Infants and Children: A Developmental Guide. McInnes, J.M. and Treffry, J.A. Toronto, Ontario: University of Toronto Press. 1984.
C M
The contents of this book include: the multi-sensory deprived child, organizing a program, social and emotional development, communication, motor development, orientation and mobility, and life skills. (284 pp.) 3 copies.

The Deaf-Blind in Higher Education (Masters Paper). Macdonald, Roderick. 1977.

Deaf-Blindness: A Functional Approach. (DRAFT). St. Paul, MN: Advisory Council to the Minnesota Deaf-Blind Technical Assistance Project.
(86 pp.)

The Deaf-Blind "Rubella" Child. Robbins, Nan and Stenquist, Gertrude. Watertown, MA: Perkins School for the Blind. 1967.

Deaf- Blind Success Stories. Smith, Anne Warren (Ed.) Monmouth, OR: Teaching Research Assistance to Children and Youth Experiencing Sensory Impairments. 1994.
Paper including sample success stories taken from contributors across the U.S. (20 pp.).

Deaf Students and the School-to-Work Transition. Allen, T.E., Rawlings, B.W., and Schildroth, A. N. Baltimore, MD: Paul H. Brookes Publishing Co. 1989.
D/HH T
This text discusses laws and regulations, reviews available services, examines existing relationships between educational programs and vocational rehabilitation agencies, and provides parents' perspectives on the transition process for individuals who are hearing impaired. (253 pp.).

Delivering Effective Instruction to Students with Deaf-Blindness and/or Other Severe Disabilities. Raleigh, NC: Exceptional Children Support Team, North Carolina Department of Public Instruction. 1995.
(206 pp.)

Developing Individualized Transition Services for Students with Deaf-Blindness (conference manual). Helen Keller National Center Technical Assistance Center. 1993.
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Conference manual from the national project supporting the transition of youth with deaf-blindness held June 24-26, 1993.

Developing Consistent and Effective Total Communication In the Home. Watkins, Susan. Logan, UT: SKI*HI Outreach, Utah State University. 1982.
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This text contains a rationale and description for the development of a total communication system in the home. (153 pp.)

Developing Personal Safety Skills in Children with Disabilities. Briggs, Freda. Baltimore, MD: Paul H. Brookes,Publishing Co. 1995.
(214 pp.)

A Developmental Profile for Use With the Deaf-Blind. Brantford, Ontario, Canada: John McInnes & Associates.
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(50 pp.)

Developmental Programming for Infants and Young Children: Volume 1. Schafer, D. Sue and Moersch, Martha S. (Eds.). Ann Arbor, MI: University of Michigan Press. 1981.
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Developmental Programming for Infants and Young Children: Volume 2. Schafer, D. Sue and Moersch, Martha S. (Eds.). Ann Arbor, MI: University of Michigan Press. 1981.
A DD EC S 2 copies.

Developmental Programming for Infants and Young Children: Volume 3.. Schafer, D. Sue and Moersch, Martha S. (Eds.). Ann Arbor, MI: University of Michigan Press. 1981.
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1 copy.

The Development of Social Skills by Blind and Visually Impaired Students: Exploratory Studies and Strategies. Sacks, Sharon Zell, Kekelis, Linda S., and Gaylord-Ross, Robert J. New York, NY: American Foundation for the Blind. 1992.
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(214 pp.)

Dimensions : Visually Impaired Persons with Multiple Disabilities. New York, NY: American Foundation for the Blind. 1989.
B/VI DD
Selected papers from the Journal of Visual Impairment and Blindness. (107 pp.)

Directory of Camps for Blind and Visually Impaired Children, Youth, and Adults. New York, NY: American Foundation for the Blind. 1989.
B/VI
(35 pp.)

Directory/Early Childhood Services

Directory of Funding Resources for Assistive Technology in Minnesota. St. Paul, MN: STAR Program, Minnesota Governor's Advisory Council on Technology for People with Disabilities.
(30 pp.)

Disability and the Family: A Guide to Decisions for Adulthood. Turnbull III, H.R., Turnbull, A.P., Bronicki, G.J., Summers, J.A., and Roeder-Gordon, C. Baltimore, MD: Paul H. Brookes Publishing Co. 1989.
F T
This text introduces guildelines for making plans that are legally and financially effective, that consider real-life choices and preferences, and that take into account the social, leisure, residential, and vocational options that can help ensure a desired quality of life for persons with disabilities and their families. (417 pp.) 2 copies.

Early Childhood Screening.
A EC

Early Focus: Working with Young Blind and Visually Impaired Children and Their Families. Pogrund, Rona L., Fazzi, Diane L., and Lampert, Jessica S. (Eds.). New York, NY: American Foundation for the Blind. 1992.
B/VI EC F

Early Intervention: Building Blocks for the Future. Trimbach, Kathy, Abderholden, Sue, and Grykiewicz, Karen. Minneapolis, MN: ARC Minnesota. 1990.
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Early Intervention for Infants and Children with Handicaps: An Empirical Base. Odom, S.L. and Karnes, M.B. Baltimore, MD: Paul H. Brookes Publishing Co. 1988.

A DD EC

This text looks at assessment, research, research evaluation and intervention in early childhood special education. (309 pp.)

Early Intervention with American Indian Families: An Annotated Bibliography. Malach, Randi, Segel, Norman, and Thomas, Trish (Eds.). Bernalillo, NM: Southwest Communication Resources, Inc.

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Early Learning Step by Step: Children with Vision Impairment and Multiple Disabilities. Nielson, Lille. Copenhagen, Denmark: SIKON. 1993.

B/VI DD EC

Early Stimulation Manual for Parents of Deaf Infants. Peace Corps.

D/HH EC F

Educational Approaches for Visually Impaired Children. Nielson, Lille. Copenhagen, Denmark: SIKON. 1992.

B/VI

Educating Children with Multiple Disabilities: A Transdisciplinary Approach (3rd Edition). Orelove, Fred P. and Sobsey, Dick. Baltimore, MD: Paul H. Brookes Publishing Co. 1996.

Educational Beginnings with Deaf-Blind Children. Robbins, Nan. Watertown, MA: Perkins School for the Blind. 1960.

Educating Students Who Have Visual Impairments with other disabilities by Sacks & Silberman.

Education of Learners with Severe Handicaps: Exemplary Service Strategies. Horner, R.H., Meyer, L.H., and Fredericks, H.D (Eds.). Baltimore, MD: Paul H. Brookes Publishing Co. 1986.

DD I

This book discusses model programs, new instructional technology, and recent advances in curriculum development that have been successfully applied in integrated settings. (375 pp.)

Education of the Visually Handicapped. Volume XX, Number 4. Winter, 1989.

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One issue of the journal.

The Educator's Guide to AIM II. Wolf, Enid G. and Schein, Jerome D.

Effective Educational and Behavioral Programming for Severely and Profoundly Handicapped Students: A Manual for Teachers and Aides. Popovich, Dorothy. Baltimore, MD: Paul H. Brookes Publishing Co. 1981.

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The content of this text includes: curriculum planning and management, appropriate learning environments, and using least restrictive environments to eliminate inappropriate behavior and maintain appropriate behaviors. (260 pp.)

Employment Options for Young Adults with Deaf-Blindness: Philosophy, Practice, New Directions. Barrett, S. and Smith, A. (Eds.). Helen Keller National Center for Deaf-Blind Youth and Adults. 1986.
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Enhancing Interactions Between Service Providers and Individuals Who are Severely Multiply Disabled: Strategies for Developing Non-Symbolic Communication. Siegel-Causey, Ellen and Guess, Doug (Eds.).1988.
C DD
3 Copies.

The EPICS Messenger. Bernalillo, NM: Southwest Communication Resources, Inc. Newsletter of the EPICS Project (Education for Parents of Indian Children with Special Needs), a national parent training and information project, phone (800) 765-7320.

Etiologies and Characteristics of Deaf-Blindness. Wolff, Katheryn, Heller, R.N., and Kennedy, Cheryl. Monmouth, OR: TRACES Project. 1994.

Everybody Counts! A Workshop Manual to Increase Awareness of Handicapped People. Ward, M.J., Arkell, H.G., and Wise, J.H. Council for Exceptional Children. 1979.
I
This book discusses workshop planning and activities to increase the awareness of persons with handicaps. Includes an audiotape. (73 pp.)

Everyday Social Interactions, A Program for people with Disabilities, by Vivienne Riches

Excerpts from Minnesota Statutes Compulsory Instruction Health Standards Nonpublic Aids. October, 1994.
H/M

Exemplary Programs for Persons with Disabilities In Transition Supported Employment, and Parent-Professional Collaboration. Minneapolis, MN: Institute on Community Integration, College of Education, University of Minnesota. 1994.
DD F I T

Express Yourself: Communication Disabilities Need Not Be Handicaps. Johnson, Peg L. Richfield, MN: Pegijohn. 1985.
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FM Systems, Use and Troubleshooting, Martie Martin, M.S.

Factors Influencing Employment Outcomes of Legally Blind Rehabilitation Clients Who Have Hearing Impairments. Maxon, B.J., Giesen, J. Martin, and Ford, Kevin. MS: Rehabilitation Research and Training Center in Blindness and Low Vision, Mississippi State University. 1986.
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A Families Guide to Advocacy. Minneapolis, MN: ARC Minnesota.
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Family-Centered Care for Children with Special Health Care Needs. Shelton, Terry L., Jeppson, Elizabeth S., and Johnson, Beverly H. Washington, DC: Association for the Care of Children's Health. 1987.
F H/M

Family-Centered Early Intervention with Infants & Toddlers: Innovative Cross-Disciplinary Approaches. Brown, Wesley, Thurman, S. Kenneth, and Pearl, Lynda F. Baltimore, MD: Paul H. Brookes Publishing Co. 1993.
EC F

Flexible Pacing for Able Learners. Daniel, N. and Cox, J. Reston, VA: Council for Exceptional Children. 1988.
The content of this text includes: introduction to flexible pacing; flexible pacing at the elementary, secondary, and all levels; cooperative programs; selected features of flexible pacing; and toward improved instruction for mathematically able students. (119 pp.) 2 copies.

Foundations of Education for Blind and Visually Handicapped Children and Youth: Theory and Practice. Scholl, G.T. (Ed.). New York, NY: American Foundation for the Blind, Inc. 1986.
B/VI
This text discusses components of a quality educational program and special curriculum considerations for blind and visually handicapped children and youth. (509 pp.) 2 copies.

Functional Vision: "Learning to Look"
B/VI

Future Is In Our Hands: A Resource Manual for Parents of Children with Disabilities.
F

GA and SK Etiquette: Guidelines for Telecommunications in the Deaf Community. Cagle, Sharon and Cagle, Keith. Bowling Green, OH: Bowling Green Press, Inc,. 1991.
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The Gallery: A Catalog of Educational Resources (Transition Resources). St. Paul, MN: Minnesota Educational Services. 1995.
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Games for People with Sensory Impairments: Strategies for Including Individuals of All Ages Lauren J. Lieberman & Jim F. Cowart

Government Policies and the Disabled in American Indian Communities by Jennie R. Joe, Ph.D. and Carol S. Locust, Ph.D. - Native American Research and Training Center, The University of Arizona, Tucson, Arizona.

Graphic Art Materials Reference Manual. Paramus, NJ: Letraset. 1984.

(Grief and Loss Packet)
F
Assorted resources related to the grief and loss families experience when a family member is a child with disabilities.

Grow Deep: Not Just Tall. Clark, Karen Kaiser. St. Paul, MN: The Center for Executive Planning, Inc. 1984.

A Guide for Parent Involvement. St. Paul, MN: Minnesota Department of Education.
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A Guide for Students Who Are Deafblind considering college.

A Guide to Helping Young Children with Visual Impairments Make Better Use of Their Vision, Book 2. Topor, Irene, Bailey, Brent R., and Houghton, Joan. IN: Blumberg Center for Interdisciplinary Studies in Special Education, Indiana State University. 1995.
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A Guide to Planning and Support for Individuals Who Are Deafblind/John M. McInnes

Guidelines for Education of Deaf and Hard of Hearing Learners. Little Canada, MN: Minnesota Educational Services. 1995.
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Guidelines from the Minnesota Department of Children, Families, and Learning.

Guidelines for Working and Playing with Deaf-Blind People. Smith, Theresa B. 1992.
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Guidelines: Practical Tips for Working and Socializing with Deafblind People by Theresa B. Smith

The Hadley School for the Blind: Course Catalog. Winnetka, IL.
B/VI
Tuition-free distance education.

Handbook for the State Recommended Individual Education Plan. St. Paul, MN: Unique Learner Needs Section, Minnesota Department of Education.

Handbook IEP

Hand in Hand: Essentials of Communication and Orientation and Mobility for Your Students Who Are Deaf-Blind-A Trainers Manual. Prickett, J. G., Joffee, E., Welch, T.R., and Huebner, K.M.(Eds.). New York, NY: AFB Press, American Foundation for the Blind. 1995.
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Hand in Hand: Essentials of Communication and Orientation and Mobility for Your Students Who Are Deaf-Blind-Volume I Heubner, K.M., Prickett, J.G., Welch, T.R., and Joffee, E. (Eds.). New York, NY: AFB Press, American Foundation for the Blind. 1995.
C M
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Hand in Hand: Essentials of Communication and Orientation and Mobility for Your Students Who Are Deaf-Blind-Volume II Heubner, K.M., Prickett, J.G., Welch, T.R., and Joffee, E. (Eds.). New York, NY: AFB Press, American Foundation for the Blind. 1995.
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2 copies.

Hand in Hand: Selected Reprints and Annotated Bibliography on Working with Students Who Are Deaf-Blind. Heubner, K.M., Prickett, J.G., Welch, T.R., and Joffee, E. (Eds.). New York, NY: AFB Press, American Foundation for the Blind. 1995.
3 copies.

Health Care Issues for Children with Special Health Needs and Disabilities: A Manual for Parent Trainers. TAPP, Federation for Children with Special Needs, Boston, MA and the Center on Human Policy, Syracuse University, Syracuse, NY. 1987.
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2 copies.

Hearing-Impairment in Children. Bess, F.H. (Ed.). Parkton, MD: York Press, Inc. 1988.
A D/HH
This book reviews etiology, assessment, and management of hearing-impairment in children. It emphasizes a multidisciplinary approach representing audiology, speech/language pathology, otolaryngology, pediatric, genetic, psychology, counseling, and educational services for hearing impaired children. (450 pp.) 2 copies.

HELP: Hawaii Early Learning Profile Activity Guide. Furuno, S., O'Reilly, K.A., Hosaka, C.M., Inatsuka, T., Allman, T.L., and Zeisloft, B. Palo Alto, CA: VORT Corporation. 1979.
A EC S
Developed by the Enrichment Project for Handicapped Infants.

Helping the Visually Impaired Child with Developmental Problems: Effective Practice in Home, School and Community. Rogow, S.M. New York, NY: Teachers College Press. 1988.
B/VI I
This book identifies and portrays the consequences of combined visual, physical, and neurological handicaps; outlines research; and explores approaches to effective intervention and curricula with an emphasis on community integration. (201 pp.)

Helping Young Children With Visual Impairments Make Use of Their Vision. Downing, June and Bailey, Brent R. Indiana State University, IN: Blumberg Center for Interdisciplinary Studies in Special Education. 1993.
B/VI EC

Holiday Book, signed english book for children, 47 pages.

Horticulture Program: Perkins School for the Blind. Watertown, MA: Perkins School for the Blind.
B/VI

How to Get Quality Care for a Child w/ Special Needs/Special Needs, Special Solutions

How to Get Services by Being Assertive. Des Jardins, Charlotte. Chicago, IL: Family Resource on Disabilities. 1993.
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How to Organize an Effective Parent/Advocacy Group and Move Bureaucracies. Des Jardins, Charlotte. Chicago, IL: Family Resource on Disabilities. 1993.
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How To Provide for Their Future. ARC, 1989.

Identifying the Needs of Drug-Affected Children: Public Policy Issues. (OSAP Prevention Monograph-11). Rockville, MD: U.S. Department of Health and Human Services. 1992.

IEP Handbook.

The Illinois Deaf-Blind Project: Post Educational Living & Working Projections For Youth 16-21. Glen Ellyn, IL: Illinois Deaf-Blind Service Center and School.
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Improving State Technical Assistance Programs. Chapel Hill, NC: The Technical Assistance Personnel Preparation System. 1986.

Independence Training for Visually Handicapped Children. Tooze, Doris. Baltimore, MD: University Park Press. 1981.
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Independence Without Sight or Sound: Suggestions for Practitioners Working with Deaf-Blind Adults. Sauerburger, Dona. New York, NY: American Foundation for the Blind. 1993.
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Individualized Assessment and Treatment for Autistic and Developmentally Disabled Children: Volume II-Teaching Strategies for Parents and Professionals. Schopler, E., Reichler, R.J., and Lansing, M. Austin, TX: PRO-ED. 1980.
A DD F
This text provides information on devising teaching strategies and provides illustrations of issues in programming. (256 pp.)

Individuals with Disabilities Education Act: Transition Requirements--A Guide for States, Districts, Schools, and Families. Stillwater, OK: National Clearinghouse of Rehabilitation Training Materials. 1996.
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Infants and Young Children with Special Needs: A Developmental and Ecological Approach. Thurman, S. K., and Wilderstrom, A.H. Baltimore, MD: Paul H. Brookes Publishing Co., 1990.
DD EC
This book gives a thorough examination of major theories of development and their relation to intervention. It addresses typical and atypical development in cognition, language, social, and motor function areas, all with an ecological perspective. For infants and young children birth to six years whose developmental patterns lie outside the typical range or whose environments and social settings place them at risk. (354 pp.)

Innovative Program Design for Individuals with Dual Sensory Impairments. Goetz, L., Guess, D., and Stremel-Campbell, K. Baltimore, MD: Paul Brookes Publishing Co., 1987.
A C M
This book discusses nonverbal or nonsymbolic communication systems, assessment of residual sight and hearing, orientation and mobility skills, functional living skills, and the use of natural cues in teaching techniques. (366 pp.) 2 copies.

(INSITE) Assessment of Developmental Skills for Young Multihandicapped Sensory Impaired Children: An Instruction Manual for the INSITE Developmental Checklist. Morgan, Elizabeth and Watkins, Sue. Logan, UT: Ski*HI Institute, Department of Communicative Disorders, Utah State University. 1989.
A DD EC S

The INSITE Developmental Checklist: A Comprehensive Developmental Checklist for Multihandicapped Sensory Impaired Infants and Young Children Ages 0-6. Morgan, Elizabeth. Logan, UT: Ski*HI Institute, Department of Communicative Disorders, Utah State University. 1989.

A DD EC S

Developmental checklist with accompanying manual available. When ordering, ask for "Assessment of Developmental Skills for Young Multihandicapped Sensory Impaired Children" ((78 pp.)

The INSITE Developmental Checklist: A Comprehensive Developmental Checklist for Multihandicapped Sensory Impaired Infants and Young Children 0-2 Years. (short version). Morgan, Elizabeth. Logan, UT: Ski*HI Institute, Department of Communicative Disorders, Utah State University. 1989.

A DD EC S

Developmental checklist with accompanying manual available. When ordering, ask for "Assessment of Developmental Skills for Young Multihandicapped Sensory Impaired Children" (49 pp.)

The INSITE Model: A Model of Home Intervention for Infant, Toddler, and Preschool Aged Multihandicapped Sensory Impaired Children--Volume I. Watkins, Susan. (Ed.). Logan, UT: SKI*HI Institute, Department of Communicative Disorders, Utah State University. 1989.

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A home intervention program for infants, toddlers, and preschool-aged children with multiple handicaps and sensory impairments. Volume I contains overview, background, and rationale for the INSITE Model; the multihandicapped sensory impaired child and his family; support services; and home visit techniques. (397 pp.)

The INSITE Model: A Model of Home Intervention for Infant, Toddler, and Preschool Aged Multihandicapped Sensory Impaired Children--Volume II. Watkins, Susan. (Ed.). Logan, UT: SKI*HI Institute, Department of Communicative Disorders, Utah State University. 1989.

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Psychotherapy with Deaf and Hard of Hearing Persons: A Systematic Model. Harvey, M.A. Hillsdale, NJ: Lawrence Erlbaum Associates. 1989.

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This book describes how one family grappled with the challenges of parenting a daughter with a mild mental handicap. (230 pp.)

The Right to Grow: Up An Introduction to Adults with Developmental Disabilities. Summers, J. A. Baltimore, MD: Paul Brookes Publishing Co. 1986.
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This work was among the first of its kind. The contents of this book include: principles of brain function, the integrative process, levels of function of the central nervous system; sensory modalities, postural responses and related functions, disorders of posture and bilateral integration, developmental apraxia, form and space perception, tactile defensiveness and related behavioral responses, auditory-language disorders, and principles and methods of intervention. (294 pp.)

Sensory Integration and the Child. Ayers, A. Jean. Western Psychological Services. 1979.

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Sex Education: A Curriculum for the Deaf-Blind. Watertown, MA: Perkins School for the Blind. 1981.

Shared Responsibility in Educational Service Delivery to Mildly Handicapped and Other Low Achieving Students. St. Paul, MN: Minnesota Department of Education. 1990.

Shifting Patterns. St. Paul, MN: Minnesota Department of Administration, Governor's Planning Council on Developmental Disabilities. 1992.

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Signing Naturally: Student Workbook Level 1. Smith, Cheri, Lentz, Ella Mae, and Mikos, Ken. San Diego, CA: DawnSignPress. 1988.

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Signing Naturally: Student Workbook Level 2. Lentz, Ella Mae, Mikos, Ken, and Smith, Chen. San Diego, CA: DawnSignPress. 1992.

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Signs and Symbols for Children-Songbook. Musselwhite, Caroline Ramsey. Asheville, NC: Caroline Ramsey Musselwhite. 1985.

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Describes issues and methods in development of shared decision making at individual school sites. Includes ideas for training and sequence of tasks for development.

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Skill Tasks: Handbook of Prevocational Activities. Birge, Barbara, Walker, Marcia L., and Majors, Martha Moran. Watertown, MA: Perkins School for the Blind. 1984.

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Social Skills for Daily Living. Circle Pines, MN: American Guidance Service.

Conversation and friendship skills; skills for getting along with others; problem solving skills. 3 copies

Socio-Cultural Aspects of Disability: A Three-Area Survey of Disabled American Indians by Felicia Hodge, Dr. P.H.,M.P.H. and Rebecca Edmonds, B.A. Native American Research and Training Center, University of Arizona

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Speak Up for Health: A Handbook for Parents. Minneapolis, MN: PACER, Inc. 1993. By PACER Center, Inc. 1993.

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A book for preparing adolescents with chronic illness or disabilities for independence in health care.

Special Devices for Hard of Hearing, Deaf, and Deaf-Blind Persons. Hurvitz, Joel and Carmen, Richard. Boston, MA: Little, Brown and Company. 1981.

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Students with Significant Challenges: Choosing and Developing Integrated Activities in the Elementary Schools. Graham, Nan. San Francisco, CA: CIPSSI Project, San Francisco State University.

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Student Transition MEGA Conference (conference manual). The Interagency Office on Transition Services.

T

Suggestions for Modifying the Home and School Environment, A Handbook for parents and teachers of children with dual sensory impairments. Perkins School f/t Blind.

A Summary Guide to Social Security and Supplemental Security Income Work Incentives for the Disabled and Blind. Social Security Administration. 1987.

Supported Employment: A Community Implementation Guide. Bellamy, G.T., Rhodes, L.E., Mank, D.M., and Albin, J.M. Baltimore, MD: Paul H. Brookes Publishing Co. 1988.

T

This text begins with an outline of 5 goals critical to successful programs and goes on to describe employment models to consider. The authors also offer several management tools to guide the reader through important management decisions, such as those related to staff training and quality control. (287 pp.)

Supported Employment: A Step-by-Step Guide. Urbain, Cathleen. Minneapolis, MN: PACER Center, Inc. 1992.

F T

Supporting Families with a Child with a Disability-An International Outlook. Gartner, A., Lipsky, D.K., and Turnbull, A.P. Baltimore, MD: Paul H. Brookes Publishing Co.

DD F

This book looks at the developments and trends in family disability issues in nine countries. It discusses three main topics-disability, culture and the family, and the lives of families with children who have disabilities. (233 pp.)

A Survival Manual for Parents of Children with Emotional/Behavioral Disorders and Mental Health Needs. Bloomington, MN: Minnesota Association for Children's Mental Health. 1995.

A F

A Survival Manual for Parents of Children with Emotional/Behavioral Disorders and Mental Health Needs: 1996 Supplement. Bloomington, MN: Minnesota Association for Children's Mental Health. 1996.

This supplement reflects changes to the resource list in original manual that will help with appropriate provision of services for children of cultural or racial minority heritage.

A Teacher's Guide To Teaching TTY Skills. Bauer, Mary. U.S. West Communications Deaf/Deaf-Blind/ Hard of Hearing Consumer Advisory Panel.

C

(17 pp.) 2 copies.

Teaching Infants and Preschoolers with Handicaps. Bailey, Jr. D.B. and Wolery, M. Bell and Howell Company. 1984.

C DD EC S

The contents of this book include: fundamentals of early intervention, determining instructional targets, implementing direct instruction, teaching imitation, designing preschool environments, working with families, acquisition and use of sensorimotor skills, preschool cognitive skills, play and social interaction with peers, reducing the occurrence of inappropriate behaviors, acquisition and use of communication skills, and acquisition and use of self-help skills. (380 pp.)

Teaching Social Skills to Youngsters with Disabilities: A Manual for Parents. Lehr, S. and Taylor, S.J. Syracuse, NY: Center on Human Policy, Syracuse University. 1987.

DD F

Prepared for the Technical Assistance for Parent Programs (TAPP) Project

Teaching the Possibilities: Identifying Individual Transition Needs. St. Paul, MN: Minnesota Department of Education. 1993.

A T

Teaching the Possibilities: Jobs and Job Training. St. Paul, MN: Minnesota Department of Education. 1991.

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Teaching the Possibilities: Home Living. St. Paul, MN: Minnesota Department of Education. 1990.

T

Teaching the Young Child with Motor Delays: A Guide for Parents and Professionals. Hanson, Marci J. and Harris, Susan R. Austin, TX: Pro-Ed, Inc. 1986.

F

The contents of this book include: developmental issues; medical aspects of motor disabilities; early intervention; interacting and teaching; observing and recording the progress; and motor, social, cognitive, and communication activities.
(218 pp.)

Technical Assistance in Educational Settings. Clifford, R.M. and Trohanis, P.L. Columbus, OH: Ohio State University. 1980.

This text discusses planning, implementing and evaluating technical assistance delivery in schools. (105 pp.)

Their Future is in Our Hands: A Resource Manual for Parents of Children with Multiple Disabilities. Leister, Chrissy, et. al. Hattiesburg, MS: University of Southern Mississippi. 1992.

F

Third Party Reimbursement: A Manual For Health Related Services Provided to Children and Youth with Handicapping Conditions. Kreb, Roberta A. and Stevens, Cynthia R. St. Paul, MN: Minnesota Department of Education. 1990.
F H/M

Together Successfully: Creating Recreational and Educational Programs That Integrate People With and Without Disabilities. Rynders, John E. and Schleien, Stuart J. Arlington, TX: Association for Retarded Citizens of the United States. 1991. **DD I**

Topics In Early Childhood Special Education: Changing Theoretical Views and Treatment Approaches. (9:3, Fall 1989). Austin, TX: Pro-Ed, Inc. 1989.
DD EC

Topics in Early Childhood Special Education: Judgment-Based Assessment. (10:3, Fall 1990). Austin, TX: Pro-Ed, Inc. 1990.
A DD EC

Transition Conference (conference manual). Minnesota W. Central ECSU. 1994.
T
Manual from the conference in June, 1994.

Transition from School to Work: New Challenges for Youth with Severe Disabilities. Wehman, P., Moon, M.S., Everson, J.M., Wood, W., and Barcus, J.M. Baltimore, MD: Paul H. Brooks Publishing Co. 1988.
DD T

Transition Planning Guide--Community Integration: A Vision for Youth with Handicaps. St. Paul, MN: Minnesota Department of Education. 1987.
DD I T

Transition Planning Guide from Home to School & Community: McLeod County CTIC

Transition Policies, Planning, and Services: Case Study Report of Minnesota. Furney, K.S., Hasazi, S.B., and DeStefano, L. 1994.

Transition Record

Transition Services for Youths Who Are Deaf-Blind: A Best Practices Guide for Educators. Everson, Jane M. (Ed.). Sands Point, NY: The Helen Keller National Center-Technical Assistance Center. 1995.
T

TRC DB-MD Waiver Training Manual, Texas Deafblind Outreach, Texas School for the Blind and Visually Impaired for Texas Rehabilitation Commission

Understanding and Conducting Qualitative Research. Stainback, S. and Stainback, W. Washington, DC: Council for Exceptional Children. 1988.
The contents include: introduction to qualitative research, characteristics of a qualitative research study, reporting qualitative research findings, credibility of qualitative research findings, using qualitative research responsibly. (115 pp.)

Understanding Low Vision. Jose, R.T. (Ed.). New York, NY: American Foundation for the Blind. 1983.

B/VI

This book discusses the assessment of low vision; clinical services; training and instructional services. (555 pp.)

A Unit Based Curriculum: An Integrated Approach for Deaf-Blind Children. Martha M. Majors, Curriculum Coordinator, Watertown, MA: Perkins School for the Blind.

Usher's Syndrome: What It Is, How to Cope and How to Help. Duncan, E., Prickett, H.T., Finkelstein, D., Vernon, M., and Hollingsworth, T. Charles C. Thomas Publishing Co. 1988.

This book contains six in-depth interviews of individuals who have Usher Syndrome followed by discussions concerning medical aspects of Usher Syndrome, how to effectively communicate, psychological adjustment, employment and vocational rehabilitation, recreation, and social interaction. (93 pp.)

Using Computers and Speech Synthesis to Facilitate Communicative Interaction with Young and/or Severely Handicapped Children. Burkhart, L.J. 1987.

Value-Based Services for Young Adults with Deaf-Blindness (conference manual). Covert, A.M. and Carr, T.S. (Eds.). Sands Point, NY: Helen Keller National Center for Deaf-Blind Youth and Adults-Technical Assistance Center. 1988.

Report of a national conference held on March 14-16, 1988 sponsored by the Helen Keller National Center for Deaf-Blind Youth and Adults-Technical Assistance Center.and The Association for Persons with Severe Handicaps-Technical Assistance Project. (107 pp.) 2 copies.

Visions: 1994 National Conference. Berkeley: CA: Conference Recording Service. 1994.

Set of twelve audiotapes from the national conference in San Francisco on November 18-20, 1994, sponsored by The Foundation Fighting Blindness.

B/VI

VISTA (Vermont Interdependent Services Team Approach): A Guide to Coordinating Educational Support Services. Giangreco, Michael F. Baltimore, MD: Paul H. Brookes Publishing Co. 1996.

Vocational Assessment: A Guide To Improving Vocational Programs for Handicapped Youth. St. Paul, MN: Minnesota Department of Education.

T

Vocational Curriculum for Deaf-Blind Youth. Watertown, MA: Perkins School for the Blind. 1983.

I T

Vocational Education: Work Experience Program for Handicapped Youth, Instructor-Coordinator Handbook. St. Paul, MN: Minnesota Department of Education.

T

Vocational Rehabilitation and Supported Employment. Wehman, P. and Moon, M.S. Baltimore, MD: Paul H. Brookes Publishing Co. 1988.

T

This text discusses general issues related to supported employment, human resources issues, training and management, community-based programs and supported employment, critical implementation issues, and supported employment applications with different populations. (372 pp.)

We Can Do It! A Curriculum for Teaching Self-Determination. Little Canada, MN: Minnesota Educational Services. 1994.

I T

Produced by a collaboration between Wilderness Inquiry, The Institute on Community Integration at the University of Minnesota, and the Minnesota Department of Education.

Welcoming Students Who Are Deaf-Blind into Typical Classrooms: Facilitating School Participating, Learning, and Friendships. Haring, N. and Romer, L. Baltimore, MD: Paul H. Brookes Publishing Co. 1995.

I

When You Have a Visually Handicapped Child in Your Classroom: Suggestions for Teachers (2nd Edition). Torres, Iris and Corn, Anne L. New York, NY: American Foundation for the Blind. 1990.

B/VI

Where Have All The Children Gone? Clark, Karen K. St. Paul, MN: Center for Executive Planning. 1977.

Wolf Inventory of Psycholinguistic Progress (WIPP): Master Administrative Manual (and set of forms). Wolf, Enid G. NY: Three Bridge Publishers. 1982.

A

2 copies.

Working Together with Children & Families: Case Studies in Early Intervention. McWilliam, P. J. and Bailey Jr., Donald B. Baltimore, MD: Paul H. Brookes Publishing Co. 1993.

EC F

Working With Families of Young Children With Special Health Care Needs. (Monograph No. 7). Morgan, Elizabeth C. (Ed.). Logan, UT: SKI*HI Institute, Utah State University. 1993.

F H/M

Young Siblings of Children with Chronic Illnesses and Developmental Disabilities. Lobato, Debra.

F H/M

Your Visually Impaired Student: A Guide to Teachers. Scott, E.P. University Park Press. 1982.

B/VI

The contents include: impact of visual impairment on normal development; attitudes, feelings, and concerns; Kindergarten, primary, intermediate, and senior grades; social and personal development; mainstreaming; Braille; and communication aids and appliances. (210 pp.)

DEAFBLIND VIDEO LIBRARY

ALPHABETICAL TITLES

1997 MN Summer Deafblind Conference, U of MN Duluth
Presenter: Ed Gervasoni, Topic: "Communication" Tape 1 of 4 (6-19-97)

1997 MN Summer Deafblind Conference, U of MN Duluth
Presenters: Ed Gervasoni, Topic: "Communication", Pam Ryan "Psychological Assessment" Tape 2 of 4 (6-19-97)

1997 MN Summer Deafblind Conference, U of MN Duluth
Presenters: Pam Ryan, Topic: "Psychological Assessment", Nancy Evelyn "Sign Language Adaptations" Tape 3 of 4 (6-19 & 20, 1997)

1997 MN Summer Deafblind Conference, U of MN Duluth
Presenters: Nancy Evelyn "Sign Language Adaptations" continued plus final wrap. Tape 4 of 4 (6-20-97)

A Glorious Accident by Oliver Sacks (Understanding our Place in the Cosmic Puzzle)

ABC: A Basic Course in American Sign Language. Humphries, Tom, Padden, Carol, and O'Rourke, Terrence J. Silver Spring, MD: TJ Publishers.
(Tapes 1-4, 1 hr. each).

Ain't Misbehavin': Strategies for Improving the Lives of Students Who Are Deaf-Blind and Present Challenging Behavior. Austin, TX: Deaf-Blind Outreach, Texas School for the Blind and Visually Impaired.
Videotape and manual providing practical strategies for proactively avoiding interactions which challenge relationships for people involved in the lives of individuals who are deaf-blind . (16 min.)

American Indian Concepts of Health & Unwellness by Carol Locust, Ph. D., University of Arizona 1990 Video

American Sign Language Dictionary, CD Rom Tutorial, Martin L.A. Sternberg, Ed. D.

Art of Observation and **Behavior Management.** (2 taped presentations). 1994.
"Art of Observation" presented by Sandra Davenport and "Behavior Management" presented by Mary Lundeen on August 14, 1994.

Assessment and the Early Years. Evanston, IL: Altschul Group Corporation. 1991.
From The Fetal Alcohol Syndrome Series by Perennial Education. (21 min.)

The Braille Butler. Ft. Collins, CO: Office of Instructional Services, Colorado State University. 1990.
Tape# x-5740. (6 min. 44 sec.) 2 copies.

Bringing Out the Best, Encouraging Expressive Communication in Children w/Multiple Handicaps. Video 24 minutes

Children with Motor Impairments. (taped presentation).
(60 min.) 2 copies.

Communication Forms with Early School-Aged Children. (taped presentation).

Deafblind, Getting Involved: A conversation, communication and community by Theresa Smith, video

Deafblind, Overview and Introduction, Communication and Community by Theresa Smith, Video

Deafblindness and the Intervener, Video Produced at: The SKI*HI Institute, Utah State University

Deaf-Blindness: Connecting Through Communication. CNIB Deaf-Blind Services. 1990. Open captioned. (14 min.)

Deaf-Blind Culture. (taped presentation). 1994. Presented by Barry Segal in 1994.

Deaf-Blind Culture and Definitions of Deaf-Blindness. (taped presentation) 1994. Barry Segal's tape is presented and discussed along with definitions of deaf-blindness on August 5, 1994.

Do You See What I See? Philip Rock Center/Helping Young Children with Vision Impairments Develop Sight Video & Booklet

Effective Education: Adapting to include all students, Video, 18:14 minutes

Encouraging Independence Exploration and **Young Children with Deaf-Blindness: Principles and Strategies** and **Communicating with Deaf-Blind.** (3 taped presentations). With Susan Hopp and Linda Hinke, Certified Interpreter.

Feeding Infants and Young Children with Special Needs / Video and folder.

Find Inc.: Independent Living for Individuals with Deaf-blindness. Akre, John.

Fingerspelling, Expressive & Receptive Fluency. San Diego, CA: DawnSign Press. 1992. Includes a video guide by Joyce Linden Groode.

Forum on Interveners. (taped presentation). 1996. With Linda Alsop and Robbi Blaha taped on July 25, 1996. 4 sets with 2 tapes per set. Tape 1 of each set interpreted. (Tape 1/113 min., Tape 2/119 min.)

Functional Vision Learning to Look. The Blumberg Center, North Dakota Department of Education and the South Dakota Department of Education and Cultural Affairs. 1993. Close captioned. For ordering information call 1-800-438-9832. (18 min.)

Getting in Touch. Communicating with a child who is deafblind.Champaign, IL: Research Press. (19 min.) 2 copies.

Gross Motor Development in Children with CHARGE: (taped presentation). (15 min.) 2 copies

Growing Up Blind and **Deaf-Blind Panel.** (2 taped presentations). 1994.

Taped on August 4, 1994 with a panel including Ron Bondroff, John Burg, Jessica Eggert, Chris Harmon, and Mary Lundeen.

Hands On. San Francisco, CA: CIPSSI Project, San Francisco State University.

Helping Your Child Learn Choices. SD: Baker Street Productions.
(9 min. 22 sec.) 2 copies.

Helping Your Child Learn Self Control. SD: Baker Street Productions.
(11 min. 2 sec.) 2 copies.

How Do We See. Logan, Utah: HOPE, Inc.
(20 min.)

Hyper Sign, Trinity Software, An Interactive Dictionary of American Sign Language

Independent Living for Individuals with Deaf-Blindness. Minneapolis, MN: FIND, Inc.

Infant Motor Development: A Look at Phases by Kerry Goudy, OTR and Joan Winger, M.S., CCC-SLP - Video

Infantastic Lullabyes/An animated world of color, shape and song...fun and learning for baby. Video, 25 minutes

INSITE: Home Visit. Logan, UT: SKI*HI Institute, Department of Communicative Disorders, Utah State University.
(17 min. 25 sec.)

Interpreting for Deafblind Students. (taped presentation).
2 copies.

Learning American Sign Language Video. Humphries, Tom and Padden, Carol. Englewood Cliffs, NJ: Prentice-Hall, Inc. 1992.
To accompany the text by the same name.

Learning Through Play with Homemade Toys - Activities to Share with Families
Alisa Burroughs & Ashley Talmadge
This book is about Making and Using Toys to Enhance Development in Children with Sensory Impairments

Life Centered Career Education: A Competency Based Approach. Reston, VA: The Council for Exceptional Children. 2 copies.

Macintosh Boardmaker 3.0, Video including counter index.

Making Connections. (taped presentation). 1995.
Made by the Minnesota Deaf-Blind T.A. Project on July 27, 1995.

Making the Most of Early Communication. Northridge, CA: Department of Special Education, California State University. 1995.

Strategies for supporting communication with infants, toddlers, and preschoolers whose multiple disabilities include vision and hearing loss.

Making the Most of Early Communication by Deborah Chen & Pamela Haag Schachter Video & Guide, Strategies for supporting communication with infants, toddlers, and preschoolers whose multiple disabilities include vision and hearing loss

Observing and Enhancing Communications Skills/For Individuals with Multisensory Impairments, Videotape 1 & 2 and Manual

Parents Can Make a Difference. W. St. Paul, MN: Armour Productions. 1993.
From the Parent Network. Open captions. (1 min. 33 sec.)

Parents' Voices: A Few Speak for Many. Minneapolis, MN: PACER Center, Inc. 1987.
Presentations by three mothers of children with severe emotional problems. Illustrates lack of responsiveness by professionals to parents. (44 min).

Positioning for Infants and Young Children with Motor Problems / Video and Folder

The Rajin' Cajun: Usher Syndrome, The Mind Traveler: Oliver Sacks, Films for the Humanities & Sciences, Videotape

Read With Me Video Series
Sharing the Joy of Storytelling with Your Deaf Toddler, Volume 1
Stories for your deaf preschooler: "Where the Wild Things Are", "More Bunny Trouble", Set 2
Stories for your deaf child:"How the Grinch Stole Christmas!","The Wolf's Chicken Stew",Set 3
Stories for your deaf child:"Elizabeth and Larry", Set 8

Sign With Me / A Family Sign Language Curriculum
Volume 1 - Birth to three - Building Conversations - Learn signs and techniques for everyday conversations with a young deaf child.
Volume 2 - Birth to three - Building Concepts - Learn to sign abstract concepts encountered daily by deaf toddlers such as funny, soft and sticky.
Volume 3 - Birth to three - Positive Parenting - Learn signs to use for praising and effectively disciplining your young deaf child.

Regular Lives. Washington, DC: State of the Art Productions. 1988.
Video made by Syracuse University. (28 min. 33 sec.)

(The Best of) Say it with Sign. Valient Educational Videos.

Billed as the "most comprehensive video series on signing ever produced". With L.J. Solow and S.N. Solow. (2 hrs.).

The Sexuality of Your Child with Disabilities. W. St. Paul, MN: Armour Productions. 1989.
Video made by PACER Center, Inc. (58 min.).

The Sibling Experience. W. St. Paul, MN: Armour Productions. 1989.
Video made by PACER Center, Inc. (63 min.).

Signing Naturally: Student Videotext Level 1. San Diego, CA: DawnSignPress. 1988.
From the American Sign Language Series using the Functional Notional Approach. With Cheri Smith, Ella Mae Lentz, and Ken Mikos. Accompanying workbook also in library. (120 min.).

Signing Naturally: Student Videotext Level 2. San Diego, CA: DawnSignPress. 1991.
From the American Sign Language Series using the Functional Notional Approach. With Ella Mae Lentz, Ken Mikos, and Cheri Smith, . Accompanying workbook also in library. Set of 2 tapes. (120 min.per tape).

SKI*HI INSTITUTE TAPES
(Also available from the SKI*HI Institute in the text library are *Sign Language for the Family: A Sign Activity and Reference Booklet* and the *SKI*HI Home Total Communication Video Tape Program Instruction Booklet.*)

SKI*HI Helping Parents through the Mourning Process. Logan, UT: SKI*HI Institute, Utah State University.
With Dr. James C. Blair. (21 min. 39 sec.)

SKI*HI Introduction To The Tactile Communication Series. Logan, UT: SKI*HI Institute, Utah State University.
Close captioned. (32 min)

SKI*HI Coactive Sign System-Tape 1. Logan, UT: HOPE, Inc.
Lessons 1 & 2 including family and foods.

SKI*HI Coactive Sign System-Tape 2. Logan, UT: HOPE, Inc.
Lessons 3 & 4.

SKI*HI Coactive Sign System-Tape 3. Logan, UT: HOPE, Inc.
Lessons 5 & 6 including daily routines for self-care.

SKI*HI Coactive Sign System-Tape 4. Logan, UT: HOPE, Inc.
Lessons 7 & 8.

SKI*HI Coactive Sign System-Tape 5. Logan, UT: HOPE, Inc.
Lessons 9 & 10 including action words and prepositions associated with daily routines.

SKI*HI Coactive Sign System-Tape 6. Logan, UT: HOPE, Inc.
Lessons 11 & 12 including going places, visiting people, and special words for Sensory Impaired Children.

SKI*HI Coactive Sign System-Tape 7. Logan, UT: HOPE, Inc.
Units 1-8 including toys, body parts, colors, letters, numbers, home, food, being sick, getting hurt, and time.

SKI*HI Coactive Sign System-Tape 8. Logan, UT: HOPE, Inc.
Units 9-16 including prepositions, clothing, cooking, eating, pronouns, holidays, bedroom, bathroom, family, people, vehicles, places, things outside.

SKI*HI Coactive Sign System-Tape 9. Logan, UT: HOPE, Inc.
Units 17-20 including descriptors (adjectives, adverbs, articles), going to school, verbs (to be verbs, helping verbs, action).

SKI*HI Home Total Communication Video Tape Program Instruction Booklet and **Guidelines for Video Store Participation in the SKI*HI Home Total Communication Video Program.** Logan, UT: SKI*HI Institute, Utah State University. 1986.

SKI*HI Interactive Series-Topics 1, 2, & 3. Logan, UT: HOPE, Inc.

SKI*HI Interactive Series-Topics 4, 5, & 6. Logan, UT: HOPE, Inc.
Includes suggestions to encourage language development and creating a communicative environment. Close captioned. (47 min). 3 copies.

SKI*HI Interactive Series-Topics 7, 8, 9, & 10. Logan, UT: HOPE, Inc.
Includes selecting materials and activities that promote interaction, establishing guidelines for effective conversation, conveying emotions and meanings through tacticle signs, and fingerspelling. Close captioned. (49 min.)

SKI*HI Interactive Series-Topics 11 & 12. Logan, UT: HOPE, Inc.
Includes encouraging interaction with others and interpreting for the individual that is deaf-blind. Close captioned. (36 min.)

SKI*HI Interactive Series-Topics 13. Logan, UT: HOPE, Inc.
Includes perspectives from individuals who are deaf-blind. Close captioned. (1 hr. 11min.)

SKI*HI Signals and Cues Series, Topic 1, 2, 3, 4

SKI*HI Signals and Cues Series, Topic 5, 6

SKI*HI Signals and Cues Series, Topic 11, 12

SKI*HI Total Communication Video Tape Program Tape 1: Family, Lesson 1, 2, 3 Logan, UT: HOPE, Inc.

SKI*HI Total Communication Video Tape Program Tape 2: Morning Routines, Lesson 4, 5, 6 Logan, UT: HOPE, Inc.

SKI*HI Total Communication Video Tape Program Tape 3: Daily Routines. Logan, UT: HOPE, Inc.
Lessons 7-9 including changing diapers, getting a drink, and using the bathroom. (26 min. 22 sec.)

SKI*HI Total Communication Video Tape Program Tape 4: Evening Routines. Logan, UT: HOPE, Inc.
Lessons 10-12 including getting dressed for bed and storytime. (27 min. 48 sec.)

SKI*HI Total Communication Video Tape Program Tape 5: Mealtimes and Snacks. Logan, UT: HOPE, Inc.
Lessons 13-15 including food, meals, and eating. (27 min. 48 sec.)

SKI*HI Total Communication Video Tape Program Tape 6: The Body & Feelings. Logan, UT: HOPE, Inc.

Lessons 16-18 including body parts, getting hurt and being sick, and expressing feelings. (23 min. 41 sec.)

SKI*HI Total Communication Video Tape Program Tape 7: Living & Working in the Home. Logan, UT: HOPE, Inc.

Lessons 19-21 including things in the house and housework, clothing care, and meals. (30 min. 24 sec.)

SKI*HI Total Communication Video Tape Program Tape 8: Playing & Doing Things. Logan, UT: HOPE, Inc.

Lessons 22-24 including toys, playing inside and outside. (28 min. 52 sec.)

SKI*HI Total Communication Video Tape Program Tape 9: Going Somewhere. Logan, UT: HOPE, Inc.

Lessons 25-27 including traveling, visiting places, and going to school. (26 min. 49 sec.)

SKI*HI Total Communication Video Tape Program Tape 10: Putting It All Together. Logan, UT: HOPE, Inc.

Lessons 28-30 including the manual alphabet, question words, connecting words, prepositions, and affixes. (28 min 47 sec.)

SKI*HI Total Communication Video Tape Program Tape 11: People. Logan, UT: HOPE, Inc.

Lessons 31-33 including relationships, identifiers, pronouns, and community people. (30 min. 52 sec.)

SKI*HI Total Communication Video Tape Program Tape 12: The Body & Clothing. Logan, UT: HOPE, Inc.

Lessons 34-36 including body parts, sickness and injury, and clothing. (27 min. 56 sec.)

SKI*HI Total Communication Video Tape Program Tape 13: Animals. Logan, UT: HOPE, Inc. Lessons 37-39 including various animal classifications. (27 min. 59 sec.)

SKI*HI Total Communication Video Tape Program Tape 14: Food. Logan, UT: HOPE, Inc.

Lessons 40-42 including various food classifications.

SKI*HI Total Communication Video Tape Program Tape 15: Describing & Feelings. Logan, UT: HOPE, Inc.

Lessons 43-45. (25 min. 37 sec.)

SKI*HI Total Communication Video Tape Program Tape 16: Inside & Outside the House. Logan, UT: HOPE, Inc.

Lessons 46-48. (29 min. 53 sec.)

SKI*HI Total Communication Video Tape Program Tape 17: Time & When Things Happen. Logan, UT: HOPE, Inc.

Lessons 49-51 including time, days, months, seasons, holidays, celebrations. (29 min. 51 sec.)

SKI*HI Total Communication Video Tape Program Tape 18: Action & Doing Things. Logan, UT: HOPE, Inc.
Lessons 52-54. (27 min. 7 sec.)

SKI*HI Total Communication Video Tape Program Tape 19: Going Places. Logan, UT: HOPE, Inc.
Lessons 55-57. (25 min. 37 sec.)

SKI*HI Total Communication Video Tape Program Tape 20: Going to School. Logan, UT: HOPE, Inc.
Lessons 58-60. (30 min. 4 sec.)

Tactile Interpreting for Students who are Deafblind. (taped presentation). 1995.
Taped on April 1, 1995.

Tangible Symbol Systems / Rowland and Schweigert / Video and Manual

Technical Assistance Teleconference, 4/97, Dr. Sandra Davenport, Paula Knutson, Sally Prouty Marshall, MN Video

Vision Tests for Infants / Closed Captioned Video

The Way To Go. San Francisco, CA: CIPSSI Project, San Francisco State University. (18 min)

(What to Do Until) The "Learning Disabilitologist" (Arrives). Oakland, CA: Department of Pediatrics, Kaiser-Permanente Medical Center.
With Joseph H. Rosenthal, M.D., Ph.D., Director of the Learning Disabilities Clinic at Kaiser-Permanente Medical Center in Oakland, CA. Discusses learning disabilities, learning differences, learning styles; a plea for individual variation. (2 hrs.)

What Can Baby Hear, video by Deborah Chen, 30 minutes, Auditory Tests and Interventions for Infants with Multiple Disabilities

What Can Baby See, video by Deborah Chen, 30 minutes, Vision Tests and Interventions for Infants with Multiple Disabilities

When Hearing Loss and Retintis Pigmentosa Happen Together: Meeting Educational Needs. Gibsonia, PA: Distance Learning Center. 1996.
Taped on March 5, 1996.

Work Experiences of Jennifer Syler/DB Videotape/Washington H.S. Teleproductions

You and Me, about educating children who are deafblind, video, 1994

You and Me, Interpreter-Tutor, video, Volume 2

You and Me, A Manual for Volume II of the You and Me Video Series

You and Me, Communication, Volume 3, A Five Part Video Series About Educating Children Who Are Deafblind.

You and Me, Social Connections, Video, Volume 4, Includes parts 4 & 5

UNTITLED TAPED PRESENTATIONS

Transition Summer Institute. 1993.
With Sister Bernadette Wynne. Set of 2 tapes.

1993 Summer Institute.
Tape 1 with Chris Harmon on August 4, 1993. Tape 2 with Katherine Stremel on August 5, 1993 . Set of 2 tapes.

CLF Weekend. 1994.
With Marcia Cushman. Taped on May 14, 1994. Set of 2 tapes.

Deaf-Blind Weekend. 1995.
With Ilene Miner. Taped on May 13, 1995. Set of 2 tapes.

5/2000

CHARGE SYNDROME:

A MANAGEMENT MANUAL

FOR PARENTS

Edited by Meg Hefner, M.S.
and Sandra L.H. Davenport, M.D.

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E. Resources
CHARGE Syndrome Foundation, Inc.
Others

HISTORY OF CHARGE SYNDROME FOUNDATION, INC. AND THE MANUAL

The CHARGE Syndrome Foundation is incorporated in Columbia, Missouri where it has its roots. It began in 1982 with the Deaf-Blind Project in the Division of Genetics, Department of Child Health, University of Missouri, Columbia. The primary participants in the Deaf-Blind Project were Dr. Joyce Mitchell (Medical Information), Dr. Sandra Davenport (Genetics), Dr. James Thelin (Audiology), and Meg Hefner (Genetics).

Although the stated aim of the Deaf-Blind Project had to do with diagnosis of CHARGE and description of the features for the benefit of geneticists and other physicians, we all quickly recognized that families need information, too. Audiologist Dr. James Thelin was at the time working with the Quota Club of Columbia, a local service organization for business and professional women. Quota Club had a pot of money and was looking for a project to fund which had something to do with hearing loss. As a result Jim and Meg became the primary authors of CHARGE Syndrome: A Booklet for Families, with lots of input from Sandy and Joyce. Quota Club member Marion Norbury volunteered to help send out the booklets. More than ten years later it has nearly taken over her life!

In 1988 we printed the first 1,000 booklets, figuring they would last five years or so (we thought CHARGE was rare!). The typical scenario was as follows: A family would write or call and request one booklet. A week or so later, the same family would request 5-10 booklets so they could be passed out to relatives, doctors, teachers, and others who worked with their child. The booklets were gone in a year, and have gone through several more printings since then. Until now, the Booklet for Families was the best initial source of information on CHARGE available to families.

After publication and distribution of the booklet, we began to get calls and letters from families asking: "Are there any other children with CHARGE in New Jersey?" "Does anyone else have a baby with feeding problems?" "Is my child's heart defect a typical one for CHARGE?" and so on. It was clear we needed a way for families to get more information and to share their information and stories with one another.

In 1989, Marion, Sandy and Meg published the first edition of the newsletter - CHARGE Accounts. The first few years were a bit shaky with issues not always coming out on time, but it was a success with families. Marion has taken primary charge of the newsletter, with editorial assistance from Meg. It now comes out four times a year at fairly regular intervals.

In 1993, we incorporated as the CHARGE Syndrome Foundation, Inc. and held our first International CHARGE Syndrome Conference in St. Louis, Missouri. We continue holding CHARGE conferences every two years with rotating geographic locations. The Foundation now has a Board of Directors and a Medical Advisory Board.

Although the original team has dispersed geographically, (Marion is still in Columbia, Missouri; Meg is in St. Louis; Jim is in Knoxville, Tennessee; and Sandra is in Bloomington, Minnesota), we remain interested and active in CHARGE (thanks in no small part to e-mail).

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As this manual goes to print, we are preparing for our 4th International CHARGE Syndrome Conference in Houston, Texas July 25-27, 1999. In 1997 our 3rd conference which was in Boston had 410 attendees of whom 135 were children (those with CHARGE and their siblings). Our membership is over 300 and we receive anywhere from 1-20 requests for information every week - by snail mail, email and phone. Our quarterly newsletter CHARGE Accounts goes to 12 countries besides the United States. A toll-free number (800-442-7604) and our own web page (www.chargesyndrome.org) make it easy for families to contact the Foundation for information and support.

Since 1997 a very active group of families, friends and professionals have been sharing their problems, support and knowledge over the Internet through an email list (see the web page) managed by one of our parents. This manual will be available for the first time at the Houston Conference.

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CHARGE STORIES FROM PARENTS

COLOBOMA

by Tim Hartshorne

Jacob's retinal colobomas were diagnosed during his first year. The coloboma in his right eye involves the macula and accounts for significant visual loss. His right eye is also smaller than his left. The coloboma in his left eye involves only part of the macula, and he seems to receive useful vision in that eye. His acuity in both eyes is poor, especially his right eye. In addition, he has astigmatism in both eyes.

A recent, functional vision evaluation found that Jacob could detect objects in his lower, right and left visual fields, but not in his upper field. His responses to visual stimuli were consistent with poor depth perception.

Jacob wears corrective lenses. He received his first pair at under a year of age, and wore them very consistently for a few years before developing a habit of throwing them. After several years of consistent effort, his school staff managed to teach him to wear his glasses while at school. However, at home he prefers to not wear them. When he is wearing his glasses he appears to be more attentive to the world around him.

Jacob's eyes are checked twice a year. A major concern with retinal colobomas is retinal detachment. So far Jacob shows no detachment.

HEART

by Jackie Kenley

Our daughter, Laura, was born in 1985 at the University of California San Francisco hospital. On her second day of life, a heart murmur was detected by the pediatric staff. The cardiology staff was then brought in for a consult and after an echocardiogram it was determined that Laura had a major heart defect common to children with CHARGE, tetralogy of Fallot. Laura had a good birth weight and we were advised that it would be optimal for Laura to grow and be older before the heart repair. As our family was dealing with the other problems our new little girl had (such as hearing and vision loss), we were grateful that the surgery could be put off. We took Laura home and cared for her while she gained weight. She also became a loving part of our family.

At thirteen months, the cardiology team felt Laura was ready for her repair. Her brother, then 6, and sister 2, were taken to the hospital. A member of the staff explained the procedure to them and showed them where Laura would be. It was a long procedure with Laura on a heart lung machine. After the first surgery, Laura had bleeding complications which only occur in a small percent of the patients. We knew at that time that Laura often fell into the "small percent" population. She had to go back on the heart lung machine while they found the area that needed suturing. Finally Laura was taken to ICU. After surgery, it was eight days before Laura could be extubated. She was kept "knocked out" with morphine during this time. It seemed an eternity for us, but finally Laura was able to come home for a quiet recovery.

Laura has had another surgery for mitral valve repair...thirteen years later. This time, Laura quickly extubated herself (much to the staff's surprise) in the ICU. Her nurses noted she was breathing fine on her own and did not seem to be in distress! She had gone into the hospital on Thursday and came home the following Tuesday! We have all witnessed an increase in learning and overall activity since this repair.

Laura's brother and sister seemed much more emotional and concerned by the second surgery. I know that they were very relieved when it was over. We do not foresee any further heart surgeries. However, with Laura, we know there is always the possibility of needing care for her heart. Our family is thankful daily for her "well-repaired heart"!

ATRESIA OF THE CHOANAE

by Debbie Matasker

My son Michael was born in 1997. He had a very eventful birth. We knew there were problems at 33 weeks, because I had developed polyhydramnios. I had an emergency C-section because of fetal distress. The polyhydramnios was probably caused by Michael's choanal atresia.

My son was delivered, cried once and then turned blue. The neonatologist tried to pass catheters down both nares but they were completely blocked, so he was immediately intubated. A CAT scan showed bilateral bony and membranous choanal atresia. The doctors at the hospital where he was born had only seen this six times in 20 years.

At two weeks of age, Michael had his first choanal atresia repair. Stents were placed for two weeks. After one week of having the stents out, his passages closed. The ENT operated again, same result. A third operation was done, again unsuccessfully. I finally got Michael discharged from the hospital were he was born and got him to Columbia Presbyterian-Babies Hospital in NYC. The ENT there operated on him for the fourth time, placing stents that had no resemblance to the other stents. These were huge stents that even stuck out of his nose and were inserted through his upper lip. This ENT left the stents in for a long eight months. Our new ENT performs about eight choanal atresia surgeries a year and sees approximately two new kids with CHARGE a year.

The stents were removed under anesthesia by our new ENT. The other ENT removed them by clipping the sutures inside the nose, which hurt Michael. Now at 26 months, his passages are still wide open. Another ENT, Dr. Cotten in Cincinnati Children's Hospital, who has the most experience in this area, said that our ENT did an exceptional job, just as he would have done. Dr. Cotten is known worldwide for tracheal reconstruction and decannulating children in cases that were particularly difficult.

Hopefully Michael's passages will remain open; our ENT feels confident that they will. This was a major medical milestone to have surpassed.

AIRWAY MANAGEMENT

by Susan Appell

Abby, our five year old CHARGER, has swallowing dysfunction resulting in copious secretions requiring frequent suctioning, tracheomalacia, bilateral choanal atresia (repaired), sleep apnea, obstructive airway as well as other medical conditions related to CHARGE syndrome. Abby's respiratory complications resulted in repeated pneumonias, pseudomonas, and frequent respiratory infections during the first year of life. During this time, Abby's cardiologist indicated that pulmonary hypertension was evident in her echocardiogram. At 10 months old, Abby was hospitalized at Johns Hopkins Children's Center in Baltimore, Maryland, for pneumonia or pseuodomonas. Her pulmonary physician ordered a sleep study which determined Abby had sleep apnea/obstructive airway. She explained that Abby could not grow or develop properly if she was not going into the appropriate levels of sleep which allowed her body to properly oxygenate itself. We had two options which included placing a trach or using the Bi-Pap Airway Management System when Abby was sleeping. Abby has used the Bi-Pap machine from 10 months of age until today. It has been a true blessing and has allowed Abby to develop mentally and physically. After using this system, there is no longer any indication that Abby suffers from pulmonary hypertension. She is able to fight off colds and has not used an antibiotic for over three years. Her secretions remain copious and her swallowing has not improved, however, if it were not for this machine, Abby would be trached or worse she may not be alive today.

The Bi-Pap Airway Management System is exactly that - it is a machine that helps manage a person's airway when he/she has difficulty breathing due to sleep apnea/obstructive airway. It does so by blowing a predetermined amount of pressure into a person's airway when they inhale with a predetermined amount of constant air pressure when they exhale. The machine can be adapted to a person's specific needs. Our Bi-Pap is made by: Respironics Inc., Murrysville, PA, 15668-8550, 412-733-0200, FAX: 412-733-0299, 800-345-6443.

The machine itself is smaller than a computer monitor and is portable. The unit filters the air and has adjustments to control the air pressure. Air pressure adjustments are made as a result of a sleep study and then routinely checked by our medical equipment company which services our machine every three months in our home. One short piece of special tubing comes out of the main machine and attaches to the humidification chamber. The humidification chamber is a small plastic bowl that sits on a hot plate. The hot plate has a temperature control which monitors the water temperature. Another tube leaves the chamber and is attached to the mask which has little portholes to allow air to escape so that carbon dioxide levels do not build up inside the mask. The child wears a little silk bonnet around her/his head which Velcro's to the mask. There are various masks available depending on your child's needs. We have learned to prepare her machine first (turned on and distilled water poured in chamber). We put Abby's mask on and then attach tubing so that air doesn't blow in her face while attaching the bonnet. We also learned to put a piece of duoderm on the bridge of her nose to reduce the pressure from the mask to prevent a pressure sore and to place Lacrilube in her eyes to prevent any drying in case the seal around the mask loosens and air blows toward the eyes. We requested an alarm system be attached to the machine so that in case of a power outage or Abby's pressure dropped an alarm would alert us.

Abby has gone from a very sickly infant who woke up every half an hour due to coughing, to a happy and fairly healthy five year old who now looks forward to going to bed and eagerly wants her mask on. She knows that this machine helps her and it surely shows when she is able to stretch out across her double bed and relax to enjoy a sound nights sleep without interruption. When Abby is on her Bi-Pap her secretions stop and unfortunately this is the only time she gets relief.

GROWTH

by Marilyn Ogan

For two years, Kristin followed a pattern of growth that ran below but parallel to the standard growth curve at the pediatrician's office. Kristin's growth then began to level off, falling further away from the normal curve as the curve began to climb. Dr. Sandra Davenport suggested we consider an endocrinology evaluation for Kristin. We consulted a pediatric endocrinologist, who reviewed her growth pattern, evaluated Kristin's nutritional intake and energy expenditure and ran some tests. The testing involved L-DOPA stimulation and blood drawing every hour for 4-5 hours. Kristin "failed" the test and was diagnosed with sever growth hormone deficiency.

Kristin began growth hormone replacement therapy in October 1995. We have had more than three years of treatment and it seems to have helped her very much. She has responded well to the therapy. One of the first things we noticed was increased muscle tone. Kristin suddenly had more strength and endurance than before. Her development skyrocketed and she was GROWING! In the 18 months prior to testing, Kristin had grown only 1/4 inch! Kristin has now reached the stage where the faster-than-normal growth rate is declining, but she is still progressing and responding to therapy. Our endocrinologist stated that Kristin was not now experiencing tremendous growth, but he felt that without growth hormone replacement therapy, Kristin would not be growing at an acceptable rate. We have to agree.

Kristin will receive growth hormone therapy for several years, until she achieves her maximum growth potential. The only drawback to the therapy from our point of view is that it is done by injection. Kristin gets a shot five nights per week. We rotate the injections between four sites. At age six, Kristin had already reached a stage where she asked to take a "rest" from the "help-me-grow shots." She has several years of treatment remaining. I anticipate it being a big issue for the family in the near future.

If you suspect a problem or are concerned about your child's growth, seek appropriate services for diagnosis. A pediatric endocrinologist would be your best choice. Don't wait for puberty to see if the child "catches up" – that is too late for growth hormone therapy to help. As a rule of thumb, any child who is only as tall as children two or more years younger or who falls away from his or her previous growth pattern should be considered for an evaluation. And remember, children with CHARGE have lots of reasons for being small, but a few really do have growth hormone deficiency.

The Human Growth Foundation (HGF) is an excellent source for information if you have questions about growth patterns, deficiency, hormone treatment, and even ways for an individual to cope with short stature. They have multiple publications (for small fees), publish several newsletters, and offer a parent-to-parent support program.

Human Growth Foundation, 7777 Leesburg Pike, Suite 202-S, Falls Church, VA 22043
(800) 451-6434 phone (703) 883-1776 fax
http://www.medhelp.org/web/hgf.htm email: hgfound@erols.com

Another source of information is the MAGIC Foundation for Children's Growth. Their website has some marvelous information about growth hormone therapy. the Frequently Asked Questions page is very informative.

MAGIC
1327 N. Harlem Avenue
Oak Park, IL 60302
(708) 328-0808 phone

BEHAVIOR

by Ana Saruski

When my son Joel's behavior became an obstacle in school and social situations, I consulted his neurologist and tried several medications to address mainly his aggressive behavior towards his peers and sibling. This aggressive behavior included pushing, pulling hair, and kicking. Several trials of different medications failed to help improve his behavior. After seeing the negative effects of some of these medications, and reading about the potential side effects, I decided to consult a natural nutritionist, who put Joel on a sugar free diet for his behavior, and dairy free diet to improve his asthma. I immediately saw changes in his behavior, which I attributed to this diet and the combination of vitamin supplements that the nutritionist recommended. His aggressive outbursts decreased, and his teachers and therapists commented on the changes in his behavior, and felt that he was much easier to work with since he started on the diet.

Joel's diet consists of pureed foods. On a daily basis, I mix the following supplements in his food: acidophilus to prevent yeast overgrowth; amino acids for protein supplementation; B complex to improve his behavior; calcium supplement to replace the calcium he used to get in dairy products; and a natural multi-vitamin. Joel has been on this diet for nearly one year. In the past month I also added a DHA supplement called "DHA Junior," which is especially designed for children 3 and over. The DHA supplement is meant to provide fatty acids which are believed to improve brain and eye function.

EARS - DEAFNESS, and LANGUAGE

by Yuka Persico

Keith, who is now almost 10, has been trached since he was six weeks old. Because he had difficulty voicing around the trach, when he was about a year old, I decided to begin signing with him. Because I believed at that time that he had hearing but couldn't voice, I only was learning and using nouns, adjectives and verbs - children picture book language. His first signs were 'more', 'please' and 'read book'. By the time he was two he was using about twenty signs, and when he was three he had a vocabulary of over 300 signs, often stringing several words together to shape a concept. Since he was learning the signs so nicely, we were becoming quite detailed in our list of fragments to describe things.

When Keith was three his hearing was routinely tested for placement in special education preschool we found that he is profoundly hard of hearing, with a 90-120 dB loss. At that time, the infant assessment team had targeted Keith for the orthopedically handicapped classroom, as he had a trach, a gastrostomy, was not independently walking and had balance issues. I had contacted the deaf and hard of hearing (DHH) teacher because I was eager to place Keith in her class even before I knew he was deaf, because signing was his mode of expression. When the DHH teacher called me back, I had already absorbed the fact that Keith was deaf, she however had not heard the news yet. Our conversation was awkward, but revealing. She began by politely outlining for me all the reasons non-deaf children use sign. She praised the IEP presentation I had prepared for Keith and was impressed by his vocabulary and my documentation of it. She began to explain how being deaf is different than being hearing and utilizing sign when I interrupted her and said "But Keith is deaf. He has a profound hearing loss!" It seems odd to say but she was very excited about this, mostly because she is a wonderful teacher and exceptional at reinforcing language. Now she was attached to Keith and to his agenda - that he needed a complete language.

There was something in the beginning of the conversation we had, before she realized Keith was deaf, and she was talking about language and vocabulary that the green light went on in my head. I had given Keith a vocabulary, but not a language. I hadn't really thought about that difference yet. I frantically began to fill in all the blanks in my signing, trying to create whole sentences, sign all the words in a storybook, and even sign all I said to anyone. Since I was quite determined about this, it really limited my conversation for a while. I was frustrated by my limited progress, and how awkward at times it was to learn the signs from a book and use them with any conviction.

Keith started special day preschool in the orthopedically handicapped class with a full time sign language interpreter, who was a deaf education student from CSUN (California State University – Northridge). When I saw her comfortably and fluently sign a complete language, I knew this is what we needed, and I invited her to come work in our home, and just sign sign sign. She came three days a week. She signed storybooks. She sat in front of the television and signed videos. She signed when she talked to me and she patiently answered all the questions I had from literally "how do I sign this.." to questions about deafness and its culture. I put ads up at Moorpark College and CSUN, and found two more college signers to come into the home the other days. Our entire family social life was based on these wonderful students and at dinnertime our table was always full of hungry students signing signing signing. I wanted not only the direct language presented to Keith, but the richness of the passive receptive language that hearing children naturally benefit from by simply being in the room when adults are speaking. I wanted to bring Keith's language to that place of fluency that deaf children of deaf parents demonstrate.

It really took about 4 years for Keith to start signing language back. At five he was indicating his preference for certain nicknames - like any three year old might do, and at six he would dictate simple repetitive stories. At seven he began to be able to read, spell and speak intelligibly, and that's when the whole thing took off. He went from simply always doing his best, to doing his best and doing fantastically. When Keith had just turned nine, he attended Space Camp in Alabama, and won the "Right Stuff" Award, ran for Vice President of Student Council for the entire Elementary School where his DHH class is located and won, and has been on the honor roll every quarter since letter grades have been given. As much as language has been the key to his emotional independence and "coming into his own" he also seems to love language, playing with it, using it and reading it.

With language, not only could we now know our child intimately, but he could freely unleash himself upon the world with joy. It's not so much that Keith is "all caught up," because he isn't, but now his progress is no longer bittersweet and we no longer compare him to the "normal" life he would never have. He has complete ownership of his own person, just like anyone else. He has his own strengths and weaknesses just like anyone else. He has become completely equal and stands on his own without qualification or explanation.

THE INFLUENCE OF SENSORY LOSS ON DEVELOPMENT

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INPUT IMPAIRMENT

In terms of intellectual development, the most important anomalies in CHARGE involve the eyes and ears. Vision and hearing are the most important "distance" senses. Some people have estimated that 70% of what we learn is through our vision and yet language develops most quickly with hearing unless the child is born into a signing environment. The combination of hearing and vision loss has been called "dual sensory impairment" or, more simply, "deafblindness." I also like to use the term "input impaired." Most measures of intelligence rely on output, i.e. how well the children use their hands, walk, talk, etc. From these outputs, an estimate of mental processing is reached. However, if input is inadequate, how can processing and output be optimal? As a result, the intelligence of children who are deafblind is routinely underestimated. While some children with CHARGE may have brain malformations which limit intelligence, *I believe the majority of these children have normal or near-normal abilities to process input.* However, that input has to be presented in a manner that the children can understand.

DETERMINING THE COMMUNICATION BUBBLE

Programs for those individuals who are blind/visually impaired usually count on hearing as a compensatory skill whereas those for the deaf/hard-of-hearing use vision. What happens if both are limited? Different approaches to learning must be found. Finding a program with at least some staff who understand dual sensory loss will be vital for the optimal development of these children. These staff can then teach those with more traditional backgrounds to adapt their programs. Using the sensory inventory to be published in a manual for interveners[1] can be useful for families and educators in determining the "communication bubble." This bubble is the area within which a child can comfortably communicate with another person or get input from the environment. The area will be different for each of the five senses: vision, hearing, smell, touch and taste.

VISION (also see section on Eyes)

If a coloboma is present just behind the iris, that section of retina will not function properly, creating a blind spot in the child's vision above the eyes. This is normally small and does not interfere much with seeing. If the cleft is large and involves a significant portion of the lower retina, then the blind spot may also be large. That child will not be able to see anything above the level, for instance, of the top of the eyeglasses. He/she will have to look up or tilt his/her head up in order to see what is up there. More significant is a cleft involving either the optic disk, which is the nerve coming into the eyeball, or the macula, which controls central vision. In either of these cases, the child may not be able to see objects clearly and distinctly no matter what kind of glasses are used.

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Many children with CHARGE have different vision in each eye. In addition, some of them may have amblyopia. This occurs when the vision in one eye is better than in the other or when the eye muscles are weak on one side so the child cannot focus well using both eyes. The brain suppresses the image from the poorer eye so they do not have to contend with double vision. Sometimes the children have eyes that are of unequal size as well. A smaller eye usually means that the cleft inside is larger or more significant; the vision on that side is usually worse.

The effect of these anomalies is an upper visual field cut and/or uncorrectable blurry vision. Such a visual field cut means the child will bump into object above him/her such as tables or open cupboard doors, or will have to tip the head back in order to look up at an adult (this can be hazardous if balance is not good).

If the macula is involved, the central vision may be very blurry or even absent. With central vision loss, the child will look above the target. For instance, in order to see a person's eyes, he/she would look at the forehead or hairline. Such children may be labeled autistic because they do not appear to make eye contact in addition to having poor communication skills because of deafness.

A *vision consultant* can help adapt materials to fit the vision needs. An *orientation and mobility specialist* may be needed to help the child move safely about the environment.

HEARING - Please see the section on Hearing in CHARGE for more details.

The hearing losses in CHARGE can be anything from mild to profound. Most children with profound hearing loss, however, do have better residual hearing than is recognized during the early hearing evaluations. Recent evidence shows that any kind of language program (oral/aural or sign) initiated for a deaf/hard-of-hearing infant within the first six months of life can lead to significantly improved language development.[2] This is clinical confirmation of the basic science evidence that the brain pathways for hearing (as well as other sensory modalities) do not develop if the input is not presented while the brain is developing.

BALANCE

When the vestibule of the inner ear is involved, the children are born without the balance sense that comes from the inner ear. This balance sense tells the child where his/her head is in space: up down, tipped sideways, or in a diagonal direction. When this sense is not present at birth, the child feels unsteady as he/she raises the head. If that same child does not see very well, there is very little motivation to raise the head. Therefore, these children have very delayed gross motor development. They prefer to keep their bodies flat on the ground or in a stable condition. They may roll to where they want to be, combat crawl, crawl with their head down on the ground (a 5-point crawl), or sit and bounce forward or backwards to where they want to be. When they get up and walk, they hang on for longer periods of time and they also keep their feet wider apart. When both vision and hearing are affected, the average age of walking in CHARGE is 3-4 years. Unsteadiness may persist into adulthood if vision is significantly impaired. The CHARGE Syndrome Foundation has a videotape of children with inner ear balance problems which shows gross motor development from infants to young adults. Children learn to trust their muscles and joints and do learn to walk and run, though on a delayed schedule.

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TONE

Many children are also reported to have low muscle tone (hypotonia). In some cases, the upper body is more involved than the lower body. This has not been well studied. However, we think that the muscles themselves are normal and that the major problem is muscle control from the nervous system.

OTHER SENSES

Touch becomes the major means of input if vision and hearing are limited. This can be with the hands, feet (preferred in some cases), face and tongue. Young children routinely mouth objects. This becomes socially unacceptable in older children and, yet, mouthing objects is a legitimate way of getting further information when other input is not available. Most children with CHARGE have a good sense of light touch. Pain sensation, however, is often altered. Many have very high thresholds for pain. Since they do not experience pain normally, they may inflict pain on others without realizing what that pain is. This can become a social problem.

Smell is frequently diminished or absent in CHARGE. The consequences are important for nutrition since smell constitutes the major part of flavor. When your nose is stuffed up from a cold, food does not taste as good. In addition, children may not understand why others move away from them when they pass gas or, as teenagers, when they take off their shoes or have just come in from vigorous exercise. They simply do not smell what others do.

Taste is probably normal but this has not been investigated. Taste buds detect only four qualities: salty and some sweet on the front of the tongue, sour on the sides and bitter in the back. Therefore, children without sense of smell may be more likely to prefer salty and spicy foods. Interestingly sweetness is not as prominent when smell is absent.

OTHER FACTORS

The child who has spent many days, weeks, or even years in the hospital does not have the normal experiences developmentally. If he/she cannot hear well, see well, or balance properly, then all phases of development are delayed. This includes gross motor, language, personal-social skills, and even fine motor skills. Understanding what is happening in the eyes and ears is, therefore, a very important task, especially in the first year of life. This rarely happens, however, because the doctors and families are more concerned with the structural problems with the nose, the palate, the heart, the kidneys, the esophagus, etc. Filling out a Developmental Timeline can be helpful in showing the interaction of the many medical concerns with developmental milestones.[3]

SUMMARY

In the long run, the measure of intelligence and the ability to develop into an independent and contributing adult depends on the level of communication that can be achieved. If a child cannot tell you how he/she thinks, you cannot measure his/her intelligence. Routinely, therefore, tests of intelligence conclude that children with CHARGE are retarded. In some cases, certainly, children have difficulty with cognition and could be called mentally retarded. In most cases, however, it is my contention that children are developmentally delayed to a very

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significant degree, but we cannot accurately measure what their cognition is. As we learn more about adolescents and young adults, it appears that some of them have significant learning disabilities. Exactly what these learning disabilities are just now being studied.

It is important to understand the interaction of the multiple anomalies present in CHARGE. From a functional point of view, it is crucial to find a way of establishing a formal communication system, both at home and at school, so these children can reach their maximum potentials.

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Introduction to Medical Aspects and Management of CHARGE Syndrome:

What this section contains, and how to use it.

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The Medical section of the CHARGE Syndrome Management Manual is divided into multiple sub-sections according to areas of the body. We intend for these to be useful both from the point of view of the families and of the professionals who work with your children. Each sub-section is has two parts: a DOCTOR section and a PARENT section. You may want to make copies of the DOCTOR pages for the specialists working with your child. Also make copies of the **OVERVIEW** page (in the INTRODUCTION section at the front of the Manual) to give to each specialist. DO NOT GIVE OUT THE ORIGINALS - KEEP THOSE IN THE MANUAL

DOCTOR section: Each section contains a short summary intended for physicians. As you may already know, most specialists do not want or need to know everything about CHARGE. They are interested in the features of CHARGE within their specialty and other features which may have an impact on how the child is treated. We have attempted to cover the range of severity of features found in CHARGE, how they may be different in CHARGE than in other children, and other factors which may need to be considered, including cautions about care for children with CHARGE. For example, it is important for the cardiologist to know about tracheomalacia and anesthesia risks before planning surgery. The DOCTOR sections are written in standard medical jargon and include medical references. We have tried to include all of the medical terms n the glossary.

PARENT section: In each PARENT section, we have tried to start out by describing the normal structure and function of the area of the body covered. Then we list some of the most common problems or abnormalities often seen in CHARGE, how the problems may be diagnosed, what tests may be done, and some of the management issues. Combined effects of multiple problems may be discussed or you may be referred to another section for the discussion.

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MEDICAL VISIT PAD / RECORD KEEPING

The typical child with CHARGE is followed by an average of 17 different medical specialists and will have more than 20 surgical procedures before he or she is 10 years old. Although the ideal would be to have a Patient Care Coordinator for your child, in reality it is the parents (most often the mother) who takes on this role. Organized record-keeping will help keep track of what is going on, aid in coordination of appointments, and facilitate communication among all concerned (parents, physicians, and other specialists).

Organization of Medical Records is an important skill for you to learn, if you haven't already. To help with this, we have designed a Medical Visit sheet. Your manual includes a separate pad containing about 50 of these sheets. Take a moment to look this over. The purpose of the visit pad is to help you keep track of office visits. An example of a completed sheet follows this description.

The top of each sheet has a place for your child's name and date of birth. The date of the appointment, name of the doctor being seen and his or her specialty should be completed, as well as where (name of hospital or clinic) the visit took place. Before each appointment, think about what you hope to get out of it. Jot down any questions you have on the upper section. During the visit, be sure your questions are addressed.

Keep the pad with you during your visit and make notes about what you are told. You may want to ask the doctor to write a short summary on the sheet for you. If any tests are done at the visit, make a note of them and when you should expect to hear any results or findings from the tests. Be sure write down your next appointment and any referrals made by this doctor.

The sheets have holes punched in them so you can keep them in a three-ring binder. You may want to keep your child's records/notes in your Manual or establish a separate binder for records. You may decide to keep all records in chronological order, or establish separate sections for each specialist.

Some parents have found it helpful to keep a master list where they list every visit in chronological order. You will be amazed at the number of appointments and the variety of specialists you and your child see. Keeping such a list can also help coordinate appointments and even medical procedures. For example, if your child needs an other set of PE tubes in her ears, the ophthalmologist may want to take advantage of her being under anesthesia to get a better look at her retina at the same time. This will take advantage of one general anesthesia to perform multiple tasks.

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CHARGE Syndrome Medical Visit Child _____

Birth Date _____

Date of Visit:	Next Visit: Time:
Specialist:	Referral to:
Specialty:	Date/Time:
Place:	Place:
Our questions: what we would like to discuss at this visit:	
Information discussed at this visit:	

_____APPOINTMENTS from ____/____/___ to ____/_____/____

Date	MD	Specialty	What was done	RTC

*Return To Clinic, e.g. 3 mo, 6 mo, 1 yr

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DIAGNOSIS, GENETICS AND PRENATAL DIAGNOSIS IN CHARGE

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HISTORY OF CHARGE ASSOCIATION AND CHARGE SYNDROME

The collection of features which came to be known as CHARGE was first recognized by Dr. Bryan Hall, who has been collecting information about choanal atresia and related anomalies since 1968. He saw a pattern emerging in children with choanal atresia and multiple anomalies and first published his findings in 1979. In 1981, Drs. Pagon, Graham, Zonana, and Young published a series of patients with similar findings and coined the acronym "CHARGE" as an easy way to remember the condition. The name is easy to remember and catchy. Unfortunately, it does not adequately cover some of the most important features seen in CHARGE syndrome.

Many physicians are still using the 1981 criteria (below) to make or rule out the diagnosis. Revised diagnostic guidelines were published by the CHARGE Syndrome Foundation Medical Advisory Board in 1998 (Blake, et al.) The patterns of defects in CHARGE syndrome can still be difficult to diagnose, even by specialists.

Syndrome or Association?

A "syndrome" is a recognizable pattern of birth defects or malformations, typically with one recognized cause (single gene or chromosome abnormality, for example). An "association" is a nonrandom collection of birth defects which is less specific than a syndrome. Until the cause(s) of CHARGE are identified, the debate about CHARGE syndrome vs. association is likely to continue in the medical genetics community. Those of us who have been most closely involved with CHARGE over the years (and some others, see Lubinsky) feel "syndrome" is a better fit for CHARGE than "association."

As with any condition, the most involved, most severely affected cases are more likely to come to medical attention and be diagnosed most easily. This means the severity of the condition may be over-estimated and the frequency underestimated (because milder cases are not yet recognized and counted). As we get better at diagnosing the milder cases, we must also revise the incidence and prognosis to better fit the entire spectrum.

ORIGINAL "CHARGE" FEATURES

- C - Coloboma of the eye
- H - Heart defects
- A - Atresia of the choanae
- R - Retardation of growth and/or development
- G - Genitourinary anomalies
- E - Ear anomalies and/or deafness

The diagnostic criteria set out in 1981 required that 4 of the 6 "CHARGE" features be present to make a definite diagnosis. However, even the 1981 paper which coined the term "CHARGE" recognized these criteria were preliminary and that the acronym did not cover all the significant findings (e.g. facial palsy) in these children.

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REVISED CHARGE DIAGNOSTIC CRITERIA (1998)

The revised diagnostic criteria take into account the fact that there are several features which are extremely common in CHARGE but very rare in other conditions (Major Diagnostic Criteria, Table 1) and features which are common in CHARGE but are also seen in several other conditions (Minor Diagnostic Criteria, Table 2). In addition, children with CHARGE may have a variety of other features (Common Findings, Table 3) which may not be particular helpful in making a diagnosis, but which can mean a lot to the family and medical community in terms of management.

A diagnosis of CHARGE should be considered in any newborn with a "Major" criterion (coloboma, choanal atresia, facial palsy, or classic CHARGE ear [Figure *1]) in combination with any other significant birth defect.

As of this writing, there is no specific test that can be done which will definitively diagnose or rule out CHARGE. CHARGE syndrome is a clinical diagnosis. That is, it is made based on physical findings along with the best judgement of the Medical Geneticist . A diagnosis of CHARGE should be made or confirmed by a Medical Geneticist who is familiar with CHARGE.

CHARGE SYNDROME: MAJOR DIAGNOSTIC CRITERIA

Features seen commonly in CHARGE, rarely in other conditions

CRITERION	INCLUDES	FREQUENCY
Coloboma	Coloboma of iris, retina, choroid, or disc Microphthalmia, anophthalmia	80 - 90%
Choanal Atresia	Unilateral (UL) or bilateral (BL); Bony or membranous; Stenosis or atresia	50 - 60%
Cranial Nerve Dysfunction	I: lack of smell VII: facial palsy (UL or BL) VIII: sensorineural hearing loss or vestibular problems IX/X: swallowing dysfunction	Frequent 40%+ 70-85% 70 - 90%
Characteristic CHARGE Ear Fig 1 ++	External Ear: Short, wide ear with little or no lobe, snipped off helix, prominent antihelix discontinuous with tragus, triangular concha, decreased cartilage, asymmetric, often protruding laterally Middle ear: abnormalities of stapes, absent stapedius tendon, cochlear anomalies	90% ?90%

Tables modified from Blake, et al., 1998, with permission

++The external ear abnormalities can be so specific as to suggest a diagnosis of CHARGE based on the ears alone.

*** see glossary and/or other medical sections for definitions of medical terms and diagrams

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OVERVIEW OF CHARGE SYNDROME FOR PHYSICIANS

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DIAGNOSIS & MANAGEMENT

The acronym CHARGE was proposed in 1981 by Pagon, et al.[1] Since then much has been learned, particularly about the influence of cranial nerve anomalies on major medical crises and long-term morbidity. The revised criteria were published in a review for physicians in 1998.[2] Morbidity can be greatly diminished by understanding the interaction of these multiple anomalies. In particular, involvement of cranial nerves IX and X cause neurologic impairment of swallowing, leading to reflux and recurrent aspiration pneumonias. Addressing this problem could prevent many hospitalizations. When failure to thrive is present, one needs to take the swallowing difficulty into account along with possible lack of olfaction and the many other anomalies like heart disease which traditionally cause poor nutrition.

Each affected system is addressed in this CHARGE Parent Manual with particular caveats related to each specialty.

DEVELOPMENT

The literature states that most, if not all, of these children are mentally retarded. *That is not true.* A majority of children have vision and/or hearing loss, which together can constitute the designation of "deafblindness" even when residual hearing and vision is present. Children who are deafblind are **"input-impaired"** which means they need to learn alternative modes of communication and different ways to explore and interact with their environments. In addition many children have Mondini malformations which lead not only to hearing loss but also to vestibular dysfunction. The developmental effects are unusual motor milestones including a "five-point crawl" and delayed age of walking. Without congenital vision deficits and other anomalies, children with vestibular dysfunction (e.g. Usher Type I) walk on average at 18-24 months. Children with CHARGE who are multiply affected walk at 3-4 years. Clearly any infant who has delayed motor milestones, does not speak, and may not look you in the eye because of a macular coloboma will be considered mentally retarded. However, measuring output in such cases does not necessarily reflect true mental processing.

As more children are put into appropriate deafblind programs with adequate input, much improved output is being observed. Early diagnosis of vision and hearing loss is important but referral to appropriate educational programming is vital.

FINDINGS DESCRIBED BY THE ACRONYM

C - Coloboma (ocular)

Cleft affects the globe but not the eyelid. The coloboma can involve the iris, retina (with or without involving the macula) or disc. An upper visual field cut can be insignificant to major.

Microphthalmos or even anophthalmos can be part of the coloboma spectrum.

C - Cranial nerve anomalies

I – the olfactory nerve may be involved with arhinencephaly. Lack of smell can have a major impact on feeding and, later, on socialization.

II – the ocular nerve is usually involved only if a coloboma involves the disc

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VII - facial palsy is usually unilateral and present at birth. Facial asymmetry without facial palsy can also be seen as can asymmetric crying facies.

VIII – the acoustic nerve may possibly also be involved separate from malformations of the ear itself

IX & X – the major early problems poor or incoordinated swallowing with gastroesophageal reflux and aspiration pneumonia. These tend to improve over weeks, months or years.

H - Heart malformations

Any of the common types may be involved but tend to be of the conotruncal variety.

Vascular rings and aberrant subclavian arteries may cause tracheal compression

A - Atresia or stenosis of choanae UL or BL, bony or membranous

R - Retardation of growth and/or development

Height and weight are usually normal at birth. Loss of growth milestones in the first two years is usually associated with failure to thrive due to swallowing problems, heart disease, hospitalizations and recurrent illnesses.

Growth hormone deficiency may be present

Developmental delay is due to many causes, the most important of which are vision and hearing loss combined with vestibular dysfunction due to anomalies of the inner ear. Acute medical illnesses and hospitalizations also contribute to the delays. Mental processing can be normal.

G – Genitourinary anomalies

Male genital anomalies include small penis, hypospadias, undescended testes

Female genital anomalies include small labia

Hypothalamic hypogonadism may account for the genital anomalies and delayed or absent puberty

Genitourinary problems also involve malformations of the kidneys and ureters (tubes to the bladder)

E - Ears anomalies: outer, middle, inner

Mixed hearing loss +/- vestibular dysfunction

Canals may be narrow

Acute and chronic otitis media is common

Anomalies of the middle ear ossicles may be present

Mondini defects vary in severity

OTHER FINDINGS

Floppy cartilage in ears and trachea

TE (tracheoesophageal) fistula

Esophageal atresia

Cleft lip/palate

DiGeorge sequence with poor immune response

High pain threshold

Resistance to some forms of anesthesia

REFERENCES:

[1] Pagon RA, Graham JM, Zonana J, Young SL. Congenital heart disease and choanal atresia with multiple anomalies. J Pediatr 1981;99:223-227.

[2] Blake KD, Davenport SLH, Hall BD et al. CHARGE association: an update and review for the primary pediatrician. Clin Pediatr 1998;37:159-174.

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CHARGE SYNDROME: MINOR DIAGNOSTIC CRITERIA:
Features less specific to CHARGE and/or not consistent enough to be considered major

CRITERION	INCLUDES	FREQUENCY
Characteristic CHARGE face Fig 2	Square face, broad prominent forehead, arched eyebrows, large eyes, occasional ptosis, prominent nasal bridge with square root, small nares, prominent nasal columella, flat midface, small mouth, occasional small chin; larger chin with age. Facial asymmetry even without facial palsy	> 50%
Characteristic CHARGE hand Fig 3	Small thumb, broad palm with "hockey-stick" palmar crease, short fingers	50%
Genital hypoplasia	Males: micropenis, cryptorchidism Females: small labia Both: delayed or incomplete pubertal development	70 - 85% Frequent ?50%
Congenital heart defects	Most common: tetralogy of Fallot, VSD, AV canal, aortic arch anomalies	70 - 85% have CHD
Cleft palate or Cleft lip	Unilateral or bilateral cleft lip +/- cleft palate Isolated cleft palate, including submucous cleft palate Can even occur with choanal atresia or stenosis	20 - 30%
TEF	Tracheo-esophageal atresia or fistula Esophageal atresia	20% 15%
Middle ear	Frequent ear infections Many sets of PE tubes	>80%
Hypotonia	Upper body hypotonia, sloping shoulders	Frequent
Renal anomalies	Hydronephrosis or reflux; Horseshoe kidney; Small or absent kidney	40%
Growth deficiency	Short stature Growth hormone deficiency	Common Rare

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Page with figures:
CHARGE Ears
CHARGE Face
CHARGE Hand

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CHARGE FACE

(1)

(2) iris coloboma

(3) R facial palsy

(4) R facial palsy

(5) BL facial palsy

Square, often asymmetric face, round eyes, flat cheekbones, wide nose with broad nasal bridge, and small chin. Unilateral facial palsy increases the asymmetry. Note that (3) and (4) are different children. With age, the face gets longer and the chin larger. (8)-(10) are the same child.

(6) teen

(7) teen

(11) **Hockey-stick palmar crease**: Upper crease on palm goes between index and middle fingers

(8) baby

(9) child

(10) teen

CHARGE EARS

(12) R ear

(13) L ear

(14) R ear

(15) L ear

(16) R ear

(17) L ear

Right and left ear of three individuals. Floppy, small, wide ears with little or no lobe, often an unfolded or clipped-off appearance to the helix (outer fold), and a prominent antihelix creating a triangular choncha (center of ear, esp. 17) The two ears usually are different.

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CHARGE SYNDROME: OTHER COMMON FINDINGS:

May be important for management, but not very helpful in making diagnosis

FINDING	INCLUDES	FREQUENCY
Brain abnormalities	Microcephaly, Agenesis of corpus callosum, Dilated ventricles	Rare
Apnea	? central (brain)	Rare
Seizures		Rare
Laryngomalacia	Can result in weak cry	Frequent
Nipple anomalies	Extra, poorly formed or misplaced nipples	Rare
Floppy cartilage	Includes tracheomalacia, floppy ears	Frequent
Thymic or parathyroid hypoplasia	DiGeorge sequence without chromosome 22 deletion	Rare
Webbed neck	Often looks like sloping shoulders	Rare
Abdominal wall defects	Omphalocele Umbilical hernia	Rare 15%
Scoliosis	Younger children Older children	Common Frequent
Limb/skeletal anomalies	Absent thumb, Polydactyly (extra fingers), Split hand	Rare
Autistic-like behavior	Often noted in childhood	Occasional
Behavior problems	Often not noted until school age	Common

Tables modified from Blake, et al with permission

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1. Hall BD: Choanal atresia and associated multiple anomalies. J Pediatr. 1979(95)395-398

2. Pagon RA, Graham JM, Zonana J, Young SL: Congenital heart disease and choanal atresia with multiple anomalies. J Pediatr. 1981(99): 223-227

3. Blake KD, Davenport SLHD, Hall BD, Hefner MA, Pagon RA, Williams MS, Lin AE, Graham JM: CHARGE Association: An update and review for the primary pediatrician. Clin Pediatr. 1998(37)159-174

4. Lubinsky MS: Properties of associations: identity, nature, and clinical criteria, with a commentary on why CHARGE and Goldenhar are not associations. Am J Med Genet 1994(1)21-25

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DIFFERENTIAL DIAGNOSIS: What else can look like CHARGE?

Chromosome abnormalities

A variety of chromosome abnormalities can result in features which overlap with CHARGE. Most have different ear anomalies and facial features. The chromosome abnormalities which overlap with CHARGE may give us clues about where to look for an abnormal gene. See also the discussion below on VCF. Children with CHARGE would be expected to have normal chromosomes, including FISH for 22.

VCF (velocardiofacial)/DiGeorge/CATCH-22 syndrome

CATCH-22 is a term coined to encompass several of the features seen in this syndrome. VCF, DiGeorge and a couple of other associations were originally thought to be different disorders. Now that there is a test available, it is known that they are all part of the same spectrum.

C - cardiac (heart) defects
A - abnormal face
T - thymic hypoplasia (small or absent thymus gland)
C- cleft palate
H - hypocalcemia (low calcium), sometimes causing seizures
22 - deletion of chromosome 22q11

"DiGeorge sequence" refers to children with heart defects in combination with thymus abnormalities, including low blood calcium levels. It can be found in children with CHARGE, VCF, or as an isolated finding.

Velocardiofacial syndrome (VCF) can include DiGeorge sequence and other CHARGE-like features including palate problems, renal abnormalities, ear abnormalities and even occasionally colobomas. The majority of children with VCF and/or DiGeorge have a microdeletion of chromosome 22 (del22q11.2) which can be detected by a special lab technique called FISH (fluorescent in-situ hybridization), thus the name "CATCH-22."

The heart defects and swallowing problems seen in VCF/DiGeorge can be similar to those seen in CHARGE. However, the characteristic ears, face and hands are distinctly different (e.g. long and slender hands in VCF vs. short and broad in CHARGE; long face in VCF, square face in CHARGE). Only about 5% of children with CHARGE have DiGeorge sequence. Conversely, of all the children with DiGeorge, about 85% have a chromosome 22 deletion, 5% have CHARGE and 10% have something else. To date, we are not aware of a single individual with definite CHARGE who had a FISH test which was positive for the 22q11 deletion.

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VATER/VACTERL association

VACTERL is a an acronym for a collection of findings which overlap with CHARGE:

V - vertebral (backbone, spine) anomalies
A - anal atresia (referring to the anus, not the nose)
C - cardiac (heart) defects
TE - tracheoesophageal fistula
R - renal (kidney) anomalies
L - limb, especially lower arm bone anomalies

Vertebral anomalies, limb anomalies and anal atresia are each very common in VACTERL and rare in CHARGE. Children with VACTERL are unlikely to have any of the major diagnostic features of CHARGE listed in Table 1. Neither do they have the typical physical features (face, ears, hands) associated with CHARGE. In some cases, especially in the newborn period, VACTERL and CHARGE may be difficult to distinguish from each other due to overlapping birth defects.

All individuals with a suspected diagnosis of CHARGE should be evaluated for chromosome anomalies, VACTERL and VCF, including chromosome analysis with FISH for the 22q11 VCF/DiGeorge locus. "FISH for 22q" is a specialized test which must be specifically requested separately from routine chromosome analysis.

PAX2

PAX2 is rare condition with features which overlap with CHARGE. Individuals with PAX2 abnormalities may have colobomas, renal anomalies and hearing loss. They do not have the facial features or ear shape associated with CHARGE. To date, we are not aware of a single individual with definite CHARGE who had a PAX2 test which was positive for any abnormalities in the gene.

Retinoic acid (Accutane)

Accutane (a drug used to treat cystic acne) taken in the first two months of pregnancy has a very high chance of causing birth defects. The ear abnormalities seen in prenatal retinoic acid exposure can be very similar to the CHARGE ear. However, the other problems caused by retinoic acid are different. Accutane taken **before** pregnancy is not known to cause any birth defects.

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WHAT DOESN'T CAUSE CHARGE

Other than retinoic acid, no specific pregnancy exposures have been linked to the features seen in CHARGE. Exposures to pesticides, smoking, alcohol use, and/or other drug use do not appear to play a role. Because the organs involved in CHARGE are developing over a number of weeks of pregnancy, it is unlikely that any one single event (car accident, food poisoning, whatever) could cause CHARGE. There is almost never any history of CHARGE or CHARGE-like features in other family members.

HOW OFTEN DOES CHARGE HAPPEN?

The incidence of CHARGE at birth is estimated to be about 1 in 12,000 births. Many infants and young children with CHARGE do not survive due to the major medical complications. Many children with a milder expression of CHARGE may not be diagnosed until they are older or missed entirely. Therefore, the number of children with CHARGE in the general population is difficult to estimate.

HOW LONG DO CHILDREN WITH CHARGE LIVE - LIFE EXPECTANCY

Life expectancy is decreased in children with CHARGE. Infants with CHARGE have complex medical problems and many of them do not survive. The highest mortality is in the first three years. Infants with bilateral choanal atresia, a complex heart defect, and/or tracheo-esophageal fistula appear to have the lowest survival rates. There is a relatively high post-operative mortality with CHARGE, possibly due to reactions to anesthesia and/or breathing or aspiration problems. The more surgeries (and anesthesias) required, the greater the risks. Even beyond infancy, many children with CHARGE require multiple surgeries and are medically fragile.

Children with CHARGE are also at very high risk for aspiration (due to TE fistula, tracheomalacia, and swallowing problems), which often leads to pneumonia. There are reports in the medical literature of as many as 30-40% of children with CHARGE not surviving to five years. However, reports in the medical literature include children with the most severe cases of CHARGE. Less severely affected children are often not diagnosed in the newborn period, and may not be included in the "survival" numbers. The actual survival of all individuals with CHARGE may be higher than reports in the medical literature.

Beyond early childhood, the mortality remains higher than in children who do not have CHARGE. This may be due to a combination of factors, including residual heart defects, continued swallowing problems, anesthesia risks and general medical fragility. Because CHARGE is a relatively recently recognized syndrome, long-term life expectancy is unknown. There are many young adults with CHARGE who appear to be in good health and have relatively few remaining medical concerns.

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RECURRENCE RISK

WILL CHARGE HAPPEN AGAIN IF I HAVE ANOTHER CHILD?

For parents with one child with CHARGE, the recurrence risk is low, probably around 1-2%. There are only a handful of documented examples of more than one child with CHARGE in a family. The recurrence risk is based on surveys of over 300 families with a child with CHARGE and extensive review of the medical literature. Most of the "familial" cases reported in the older medical literature probably would not be diagnosed as CHARGE today based on the revised criteria. Even using a very loose definition of CHARGE (i.e. anyone diagnosed as CHARGE, regardless of the expertise of the person making the diagnosis, the certainty of the diagnosis or the criteria used), the recurrence risk is still only about 1-2%. Using strict diagnostic criteria, the risk would probably be even lower.

WILL CHARGE HAPPEN AGAIN TO ANYONE ELSE IN THE FAMILY?

Aunts, uncles and siblings of individuals with CHARGE probably are not at increased risk for having a child with CHARGE, although this information may change as we learn more about the cause of CHARGE.

Some individuals with CHARGE may be capable of having their own children when they become adults (many may not, due to hormone abnormalities). If CHARGE syndrome is caused by a single gene or chromosomal microdeletion, the risk to children of affected individuals could be as high as 50%. Information in this area may change very quickly, so be sure you have current information before making family planning decisions.

FUTURE PREGNANCIES/PRENATAL DIAGNOSIS

The recurrence risk is low, but it is not zero. Parents are understandably worried about attempting another pregnancy and want to know what can be done to look for signs of CHARGE during a pregnancy. **There is nothing that can be done to prevent CHARGE**. However, as much reassurance as possible during the pregnancy that things appear to be going well is helpful.

The way to get the most information about CHARGE during a pregnancy is to have a directed ultrasound to look for features associated with CHARGE. This should be a **Level II ultrasound exam** and should be performed at a **tertiary care center** by an experienced ultrasound technologist using state-of-the-art equipment. This is not a procedure that can be done in the typical obstetrician's office. The ideal would be two ultrasound exams, the first at 18-20 weeks (post LMP) and the second about a month later, at 22-24 weeks. Take along the following checklist to give the sonographer the best information possible about what to look for.

ULTRASOUND EXAM FOR FEATURES OF CHARGE SYNDROME:
THIS WILL NOT DIAGNOSE OR RULE OUT CHARGE

The ultrasound evaluation should include a **complete standard anatomic survey** with particular attention to the following:

- Amniotic fluid measurement:
  Look for polyhydramnios (excess amniotic fluid) associated (in late pregnancy) with choanal atresia, esophageal atresia or poor swallowing.
- Cardiac evaluation:
  Many centers can do a formal fetal echocardiogram. Heart defects most common in CHARGE include tetralogy of Fallot with or without AV canal, and right-sided anomalies, including VSD and aortic arch anomalies.
- Kidney:
  Any kidney anomaly can be associated with CHARGE, including hydronephrosis (excess fluid in the kidneys), small or absent kidney, horseshoe kidney, posterior urethral valves.
- Brain:
  Dilated ventricles, absence of the corpus callosum or any other structural abnormality of the brain.
- Face: cleft lip or cleft palate
- Ear: abnormal shape or placement (take along a photo of CHARGE ears)
- Genitalia: small penis in a known male fetus

Many abnormal findings would not be present early on and/or would be undetectable until later in pregnancy. Don't be shy about asking how confident they are about the accuracy and completeness of the ultrasound exam. Every exam is different and none will detect every birth defect during pregnancy. The accuracy will depend on a number of things, including how far along you are, the position of the baby, your weight (the image is not as clear when it has to travel through a lot of maternal tissue before it reaches the fetus), the quality of the equipment, and the expertise of the sonographer. What can be seen one day may not be visible another day.

Remember, **none** of the major diagnostic criteria for CHARGE (coloboma, choanal atresia, cranial nerve abnormalities, characteristic CHARGE ear) can be definitively diagnosed by prenatal ultrasound exam. Although finding evidence of some problem or potential problem through the ultrasound exam would certainly raise the suspicion of CHARGE, and can help parents be prepared for that possibility, it is not diagnostic. And remember, **a completely normal ultrasound exam cannot rule out CHARGE.**

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For the Neurologist: The Brain in CHARGE Syndrome

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Brain anomalies seen in CHARGE

Cranial nerve anomalies (70-90%)
- I - olfactory - (arhinencephaly in 42%)
- II - optic (colobomas in 80%)
- III, IV, V, VI rarely affected
- VII – facial palsy, UL or BL (43%)
- VIII - acoustic (60 - 80%+)
- IX, X glossopharyngeal/vagus (50%+) swallowing
- XI accessory - rarely affected moving head back & forth & shrugging
- XII hypoglossal - rarely affected move tongue ~~to~~ left - right

Structural anomalies (less common)
- agenesis of the corpus callosum
- arhinencephaly
- holoprosencephaly
- hydrocephalus
- cerebral dysgenesis
- Mondini defect
- hindbrain defects
  - cerebellar hypoplasia
  - Dandy-Walker malformation

Other
- seizures
- behavior abnormalities
- learning disabilities

Development: although it has been reported in the past that a majority of children with CHARGE are mentally retarded, this is not necessarily the case. It is extremely important for parents to realize that their child may have significant (even near-normal) potential, given accurate diagnosis and appropriate management of sensory deficits and other complications of CHARGE.

Diagnostic tests/referrals
- MRI of brain
- CT
- EEG (if suspect seizures)
- Evoked potentials (vision/hearing)
- Other hearing tests (otolaryngology)
- Dilated eye exam (ophthalmology)
- Developmental testing (developmental pediatrics/deafblind specialist)

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Medical treatment caveats
Possible unexpected reaction to anesthesia: Some children with CHARGE are resistant to sedation, while others are slow to recover from anesthesia. Because of risks of anesthesia, it may be appropriate to combine surgical procedures in these children.

Laryngomalacia or tracheomalacia is common in CHARGE and can result in surgical or anesthetic complications. The mortality rate in children with this combination of features is high.

Reflux, facial palsy and other cranial nerve anomalies may additionally compromise feeding in these children. There is a very high risk for repeated aspiration pneumonia.

If MRI is being considered, also consider a few more cuts to view the inner and middle ear, as there is a very high frequency of ossicular malformations and/or Mondini defect.

Non-medical management:
Cranial nerve IX, X, and VII anomalies can have an enormous impact on feeding. Feeding issues and common aspiration pneumonia are among the most important day-to-day issues for families. Often a feeding specialist can help the family cope with these issues.

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THE BRAIN IN CHARGE: PARENT INFORMATION

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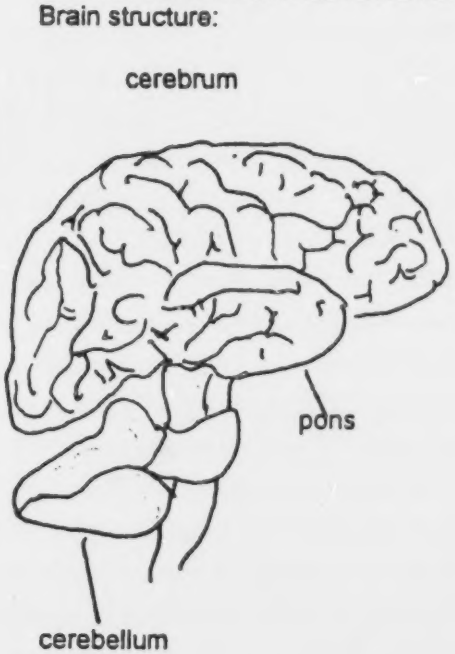
NORMAL BRAIN STRUCTURE AND FUNCTION:
Brain structure:



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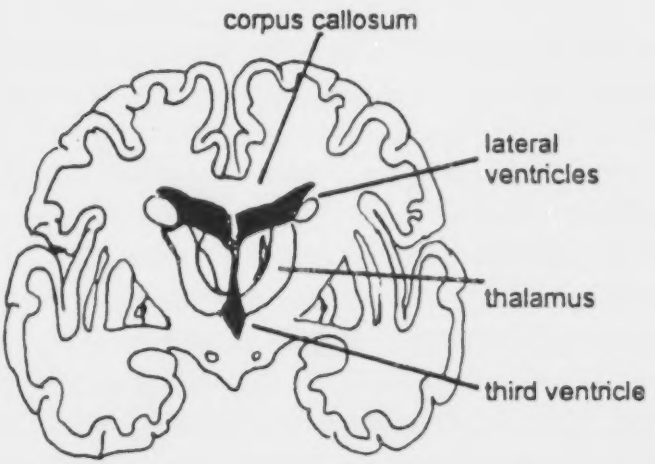

Cranial nerves: there are 12 cranial nerves, which begin in the brain and extend to structures in the head and neck. These nerves provide both motor control and sensation and include nerves involved in the senses of smell, taste, hearing, and vision, as well as movements of the eyes, face, tongue, palate, and swallowing. Some cranial nerves are also involved in the control of heart rate and movements of the gastrointestinal tract. Cranial nerve anomalies are very frequent in CHARGE.

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BRAIN ABNORMALITIES SEEN IN CHARGE

Cranial Nerve Anomalies :

Anomaly	Frequency	Tests Used	Specialist
I – olfactory (smell)	? 40%	Clinical assessment	neurologist/otolaryngologist
II – optic nerve Coloboma	80-90%	Dilated exam Visual evoked responses	Ophthalmologist
VII – facial palsy	42%, usually one sided	Clinical assessment	neurologist
VIII -- sensorineural hearing loss	60-80%	ABR Other hearing tests	pediatric audiologist otolaryngologist
IX, X swallowing and oromotor problems	50%+	Barium swallow Laryngoscopy	otolaryngologist OT, PT, speech pathology
XII – tongue	rare	Clinical assessment	neurologist

Structural brain anomalies:

A variety of structural malformations of the brain have been reported in children with CHARGE. Pretty much any brain anomaly is consistent with CHARGE, none are extremely common. A neurologist may order brain imaging such as MRI or CT scan to look for possible structural brain anomalies. Because not all children have had imaging performed, it is not possible to list the frequency of each anomaly.

1. Arhinencephaly: absence of the olfactory lobes
2. Holoprosencephaly: abnormal formation of the cerebral hemispheres.
3. Agenesis of the corpus callosum: lack of fiber tract connecting the two hemispheres.
4. Cerebral dysgenesis: abnormal formation of the cerebral cortex.
5. Hydrocephalus: increased fluid in the ventricles of the brain

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Other brain abnormalities described in CHARGE

1. Seizures: diagnosed by EEG
2. Behavior abnormalities: See DEVELOPMENT sections
3. L earning disabilities: SEE Development section

EFFECT OF ABNORMALITY ON CHILD

Cranial nerve abnormalities: cannot be surgically corrected. Only a few (hearing loss, swallowing problems) are amenable to management.

Olfactory nerve (I):
This nerve controls the sense of smell. If it is abnormal, the child's sense of smell (and therefore taste) will be absent or abnormal. This can complicate feeding problems.

Optic nerve (II):
A majority of children with CHARGE have colobomas. Optic nerve colobomas can severely affect vision. See EYE section.

Facial palsy (VII):
Droopiness or weakness of one (unilateral, UL) or both sides (bilateral, BL) of the face. This may affect feeding, with drooling on the affected side. If bilateral, there may be problems keeping food in the mouth. Facial palsy can have an effect on facial expression. In some cases, the eye on the affected side may not close completely and need to be kept moist with artificial tears. In some cases, facial nerve palsies get better over time. See FEEDING section.

Acoustic nerve (VIII):
Abnormalities of cranial nerve VIII will result in sensorineural hearing loss (see EARS AND HEARING).

Glossopharyngeal and vagus nerves (IX and X):
These nerves are involved in coordination of suck and swallow and with some of the mouth movements involved in speech. Feeding and swallowing problems are very common in children with CHARGE (see FEEDING). Many of these problems get better over time (often years), due to a combination of maturation of the nerve and use of appropriate therapies.

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Structural brain abnormalities: most are not treatable

1. Arhinencephaly will cause lack of smell. This can be difficult to diagnose, but has been described very often at autopsy in CHARGE.

2. Holoprosencephaly is a serious brain abnormality which is likely to result in significant mental retardation.

3. Agenesis of the corpus callosum is often (but not always) associated with learning disabilities and/or mental retardation.

4. Hydrocephalus can be associated with learning disabilities or mental retardation. Hydrocephalus is treated by placing a shunt.

5. Cerebral dysgenesis: many children with CHARGE have structural brain abnormalities noted on MRI or CT scan. The significance in terms of functioning is not always clear.

Other brain abnormalities described in CHARGE

1. Seizures: diagnosed by EEG, often effectively treated with medication

2. Behavior abnormalities: See DEVELOPMENT sections.

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THE EYES IN CHARGE FOR THE OPHTHALMOLOGIST

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OPHTHALMOLOGIC ANOMALIES SEEN IN CHARGE

- Coloboma (80-90%)
  - Iris
  - Retina
  - Optic nerve
  - Microphthalmia
- Facial nerve palsy (40%, unilateral >>bilateral)
- Visual acuity abnormalities (90%)
- Strabismus or amblyopia (frequent)
- Ptosis
- Cataracts
- Retinal detachment
- Photophobia (frequent)

DIAGNOSTIC TESTS

- Dilated funduscopic examination

MEDICAL CONSEQUENCES

- Colobomas of the iris typically do not affect visual acuity or visual field
- Colobomas of the retina cause visual field defects in the upper visual field. They also predispose the patient to retinal detachment.
- Colobomas of the macula and/or optic disk usually affect visual acuity significantly.
- Facial palsy can result in lack of blinking and resultant dry cornea which can lead to corneal scarring.

MEDICAL MANAGEMENT WITH CAVEATS

- Accurate description of visual acuity and visual field are of paramount importance for educational and communication purposes, particularly since most children with CHARGE have mild to profound hearing loss as well.
- Glasses (spectacles) to correct refractive error
- Tinted glasses for photophobia
- Occlusive patching for treatment of amblyopia
- Surgery for strabismus, cataracts, retinal detachment, as appropriate
- Artificial tears or gel to treat corneal exposure associated with facial palsy
- Regular (yearly) ophthalmologic evaluations to asses changes in visual acuity, refractive error, and potential for retinal detachment. Parents should be informed of the risk of retinal detachment and the importance of immediate medical assessment if there is any change in the vision status of the child.

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NON-MEDICAL MANAGEMENT ISSUES

- A diagram of the visual fields should be given to families and vision teachers or therapists so that communication programming will be optimized. The visual field may be a crescent-shaped area of the lower visual field. Some children will tilt their heads back in order to compensate to see. Accurate assessment of the most comfortable head position for viewing objects is important.

- Low vision aids such as magnifying bars, televisions and binoculars may be helpful

- Many children with CHARGE are sensitive to bright lights. Sunglasses can be very helpful in making the child more comfortable.

- Often the child appears to see better than would be predicted based on results of formal acuity and visual field testing. Many children who are legally blind function quite well visually. The parents and teachers usually can provide an excellent description of what the child can see.

- Demonstrate for parents what the vision is with best correction to help them understand what the child can and cannot see. For instance, parents frequently misunderstand that children with high myopia can see object moving at a distance when lighting and contrast are adequate, but cannot see detail clearly. In this situation, parents often have the impression that the child is not significantly visually impaired, when that is not the case.

- Most children with CHARGE have multiple anomalies, especially hearing loss. Significant vision problems combined with facial palsy, deafness, and inability to communicate may result in autistic-like behavior. However, once vision and hearing have been accurately assessed and an appropriate communication pattern established, such behaviors are often extinguished. Evaluation by a deafblind specialist (not simply a low-vision specialist) is essential.

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THE EYES IN CHARGE – PARENT INFORMATION

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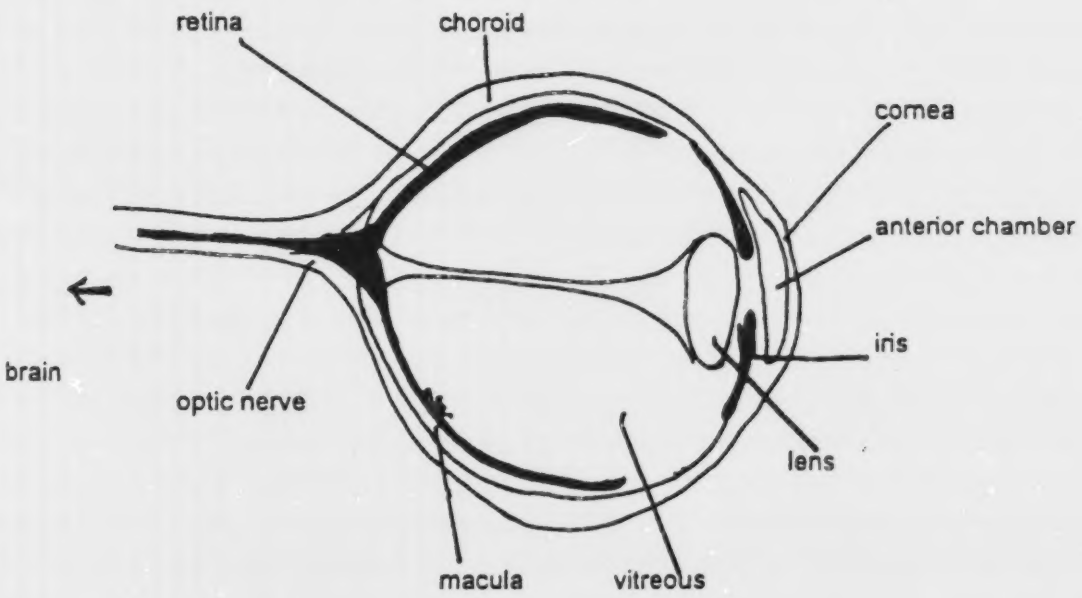
NORMAL STRUCTURE AND FUNCTION OF THE EYE

Structure: Parts of the eye



Function

- Transmission of light
  - cornea
  - aqueous humor
  - lens
  - vitreous humor
- Conversion of light to electricity: retina
- Transmission of electrical signals to the brain:
  - optic nerve (cranial nerve II)
  - optic tract
- Interpretation of electrical signals: occipital cortex and surrounding tissues (brain)

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Problem List : eye problems seen in CHARGE

Problem	Test(s)	Specialist
Coloboma of the iris (keyhole pupil)	External examination	Ophthalmologist Pediatrician/Family physician
Coloboma of retina, optic nerve	Dilated eye examination	Pediatric ophthalmologist
Visual acuity (blurriness)	Eye charts or cards	Pediatric ophthalmologist
Visual field defects (blind spots)	Dilated eye examination Visual field testing	Pediatric ophthalmologist
Retinal detachment	Dilated eye examination	Ophthalmologist - retinal specialist
Corneal exposure secondary to facial palsy	External examination	Ophthalmologist Pediatrician
Cataracts	External examiniation	Ophthalmologist
Ptosis (droopy lids)	External examination	Ophthalmologist
Strabismus or amblyopia (weak eye)	External examination	Ophthalmologist

EFFECT OF PROBLEM ON CHILD

Iris coloboma:
This does not affect vision, but may make the child more sensitive to light (photophobia).

Retinal coloboma:
This will result in large blind spots, usually in the upper field of vision, (as if the child were wearing a baseball cap). Many children with retinal colobomas prefer to be upside down and to bottom-shuffle, in part because that way they can best make use of their available visual field. .

Coloboma of the macula or optic nerve:
This often results in blurry vision as well as large blind spots. Children with extensive colobomas are often legally blind (20/200 acuity or worse). They may not look directly at objects or establish eye contact because of poor central vision.

Microphthalmia (small eye)
This can be associated with large colobomas of the retina.

Any coloboma of the retina or disk puts a child at increased risk of retinal detachment. Any sudden change in vision should be treated as a medical emergency.

Strabismus
Loss of vision can result if not corrected.

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ENDOCRINE PROBLEMS IN CHARGE FOR THE PHYSICIAN

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Although genital anomalies are common in CHARGE and make up part of the diagnostic criteria (minor criteria), there are few data on their management and outcomes, especially long-term. As a result much of the data is anecdotal, and recommendations are therefore based more on experience in groups of children with other similar anomalies.

TYPE & FREQUENCY OF GENITAL ANOMALIES IN CHARGE SYNDROME

	Frequency
Males:	
Micropenis	85%
Undescended testes	60%
Females:	
Hypoplastic labia	Very common
Delayed/arrested puberty (males and females)	Very common
Infertility	Unknown

STUDIES TO CONSIDER

It is thought that the majority of children with CHARGE have isolated hypogonadotrophic hypogonadism (i.e. pituitary gonadotropin deficiency) to account for their genital anomalies. For instance, although a number of different factors are involved in the descent of the testes and the development of the penis, it is clear that gonadotropins (mediated through testosterone) are required. Investigation is therefore aimed at:

- Assessment of gonadotropin function.
- Assessment of gonadal dysfunction arising from:
    1) Gonadotropin insufficiency
    2) Undescended testes in males

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INFANTS

Postnatally normal infants show a rise in LH, FSH and testosterone in males and estrogen in females which peaks at 8 weeks and subsides by 6 months. This "mini-puberty" offers a window of opportunity to investigate these children using baseline bloods alone in the six months of life. In males a peak serum testosterone concentration over 100ng/dl can be regarded as normal.

CHILDHOOD & ADOLESCENCE

Between the ages of 6 months and the onset of puberty the levels of gonadotropins, testosterone and estrogen remain low. At this age the following tests are recommended:

1. LHRH (GnRH) stimulation test, looking at pituitary gonadotropins (LH & FSH).

2. hCG test (in males only). Human chorionic gonadotropin – equivalent to LH, tests Leydig cell function alone. There should be a three-fold rise in testosterone following the injections; the response reflects the amount of functional testicular tissue.

3. For girls pelvic ultrasound should be used to assess the internal genitalia, and the response to therapy.

4. Where appropriate, tests of other pituitary hormones may be performed, such as baseline T4, T3, TSH, 9am cortisol, prolactin, insulin-like growth factor-1 (IGF-1) and its GH-dependent binding protein (IGF-BP3), electrolytes and plasma/urine osmolality.

5. Formal anterior and/or posterior pituitary function.

THERAPY

<u>Micropenis</u>

If the underlying cause is thought to be hypogonadotrophic hypogonadism, then the treatment is testosterone replacement. This can be given by intramuscular injection (testosterone enanthate or propionate 12.5-25mg 3-4 weekly for 3-4 doses). Topical testosterone cream 2% is also available and is administered once or twice daily for up to 3 months. Absorption may be erratic and (especially if female) the person applying the cream must ensure that they wear gloves. Some pubic hair growth may occur after testosterone administration. The most important single predictor of adult penile size appears to be the initial length of the penis.

It is unlikely that hypoplastic labia need any therapy. 118

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Cryptorchidism

The optimum timing and mode of therapy to bring down the undescended testis is contentious, even in "normal" boys. Histological changes occur in the cryptorchid testis within 1-2 years, although this must be balanced against the increased technical difficulties of surgery at younger ages. Even if the testes are not felt to have much potential for function, many surgeons would still perform orchidopexy to reduce the chances of detection of malignant change.

Hormonal therapy with hCG (human chorionic gonadotropin) may be appropriate (especially for palpable testes), and is usually given after the age of four years (with a 50% success rate). Traditionally 500-1000IU is given intramuscularly twice weekly for 5 weeks. This may also cause increased penile growth as well as producing testicular descent.

Delayed/absent puberty

MALES

- Intramuscular testosterone enanthate or propionate, 50-250mg monthly.
- Oral testosterone undecanoate 20-120mg daily.
- Patches and long-acting subcutaneous pellets have been used in hypogonadal boys with some success.

FEMALES

- Ethinyloestradiol initially 2mcg/day, increasing over approximately 2-3 years to 10mcg/day. When full pubertal progression has occurred or if there has been breakthrough bleeding then change either to low dose oral contraceptive pill or adult HRT.

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CAVEATS

- Testosterone can cause fluid retention and should be used in caution in children with heart failure.
- As hCG testing investigates the Leydig cell function of the testis, an absent response does not therefore necessarily confirm that there is no testicular function.
- hCG should not be used in girls as it can cause ovarian stimulation.
- Although relatively rare, hypopituitarism may also occur in CHARGE, and the following may indicate that further investigation of the other pituitary hormones is required:
  - Other midline defects such as clefting.
  - Conjugated hyperbilirubinemia in the neonatal period.
  - Hypoglycemia in the neonatal period.
  - Absence of pituitary or other midline structures on brain scanning.
- Animal studies have suggested that early exposure to exogenous testosterone may down- regulate the androgen receptor, leading to poor penile size in adulthood. This has not been shown in human disorders where there is early exposure to androgens.
- There are concerns regarding long-term oral testosterone therapy as this has been shown to cause liver dysfunction in some patients.
- If the testes are retained intra-abdominally it is highly unlikely that fertility can be achieved, and even if the testes are brought down then fertility is likely to be compromised. It is also recognised that the risk of malignancy is higher in those with previously cryptorchid testes.

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GENITAL PROBLEMS IN CHARGE SYNDROME for the Parent

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GENITAL ABNORMALITIES SEEN IN CHARGE

Although genital abnormalities (minor diagnostic criterion) are common in CHARGE, there is very little information in the literature describing these problems or treatment. More importantly, as children may have other potentially more serious problems (especially at birth) the genital abnormalities are often not noted or treated.

In both sexes with CHARGE the main problem seems to be an abnormality in production of the hormones (chemical messengers) from the pituitary gland (a small pea-sized gland lying underneath the brain) which control the production of sex hormones from the testicles in boys, and ovaries in girls. This can result in:

- In boys, undescended testicles (one or both) and small penis (micropenis - less than 2 cm stretched length) at birth. About three-quarters of boys with CHARGE will have micropenis. About half will have undescended testes. Both problems are due to the fact that descent of the testicles into the scrotum and growth of the penis in the last part of the pregnancy are dependent on the production of the hormone testosterone from the testicles (under the influence of pituitary hormones).

- In girls, the clitoris and labia (vaginal lips) may be smaller than usual, but this will not be as obvious as in the boys. It is very common in girls with CHARGE.

- In both sexes, there may be failure or slow progress in puberty. This will mean that the boys will not get increase in the size of their testicles and penis, and the girls will not develop breasts or begin their periods without hormone treatments. In both sexes, there will be failure of development of pubic hair.

- Infertility. The outlook for fertility in children with CHARGE is unknown.

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TESTS WHICH MAY BE RECOMMENDED

BASE BLOOD TESTS

These measure the amount of hormones in the blood, either on a single sample (base levels) or after the levels are stimulated (usually by a hormone injection). The testicles and ovaries are both quite active in the months before and after birth, and base levels may be quite helpful. From about six months of age until puberty the production of sex hormones is very low, and therefore it is often necessary to measure hormones after chemical stimulation.

- LH (luteinizing hormone) produced from the pituitary gland is responsible for the production of testosterone in boys, and for the production of estrogen and progesterone in girls.
- FSH (follicle stimulating hormone) produced from the pituitary gland is responsible for assisting in the production of sperm in the male, and ovulation in the female.
- Testosterone, the male sex hormone, produced from the testicles.
- Estrogen, the female sex hormone, produced from the ovaries: this causes breast development.
- Progesterone, another female sex hormone from the ovaries, when present along with estrogen produces menstruation.

STIMULATION TESTS

- LHRH test. In this test the production of LH and FSH from the pituitary gland is measured using luteinizing hormone releasing hormone (LHRH) as the stimulatory hormone. This is usually done over one hour, with samples taken after 0, 30 and 60 minutes after intravenous injection of LHRH. In most patients with CHARGE there is little or no increase in LH & FSH during the test.
- hCG (human chorionic gonadotropin) test. By giving several injections of a substance similar to LH over several days, this measures the ability of the testicle to produce the male hormone testosterone.

Although hCG will also stimulate hormone production in girls it may cause ovarian over-stimulation and is therefore not usually used in girls.

IMAGING

In girls ultrasound can be used to assess the size of the ovaries and uterus (womb).

In boys ultrasound may be used to assess the position of the undescended testicles, although other scans may be more accurate.

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TREATMENT

UNDESCENDED TESTICLES

If the testicles are undescended they need to be brought down. This is usually done surgically, requiring one or more operations. Ideally this should be done as early as possible, although other medical problems of CHARGE and technical difficulties of performing surgery in young babies usually means that it is performed when they are older. Alternatively, the testicles can sometimes be brought down using hCG injections (usually twice weekly over 3-6 weeks), and there is some evidence that it works best in testicles which are not completely undescended, and after 4 years of age.

MICROPENIS

A penis which at birth is less than 2cm (stretched) is defined as a micropenis. This should be treated with testosterone. This can be given as a cream applied twice daily for up to three months. As it is absorbed through the skin, the person applying it should wear gloves. Alternatively, testosterone injections can be given monthly for 3-4months. Although there have been theoretical worries that early treatment will affect the growth of the penis during puberty, this does not appear to happen. *Testosterone can cause fluid retention and should be used in caution in children with heart failure.*

The small labia in girls are not usually treated.

DELAYED PUBERTY

Although there may be worries about worsening behavioral problems during teenage years by treating with sex hormones, this must be balanced against the long-term risks of osteoporosis. The timing of treatment will take into account the normal age of puberty in boys and girls (from 11 years of age), but may be delayed to see if puberty occurs spontaneously.

GIRLS: Gradually increasing doses of estrogen in tablet form is used. Once full pubertal development has occurred, adding in progesterone either as hormone replacement therapy (HRT) or in the low-dose birth control pill (BCP) will produce periods if the uterus is of sufficient size.

BOYS: Gradually increasing doses of testosterone are given by injection, tablet or patch. In both sexes it is likely that hormone replacement will need to be given long-term.

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URINARY TRACT ANOMALIES IN CHARGE - for the Urologist

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TYPE AND FREQUENCY OF URINARY TRACT ANOMALIES IN CHARGE SYNDROME

- 20-40% have a urinary tract anomaly (1,4).

- All types of structural problems (solitary kidney, hydronephrosis, renal hypoplasia, duplex kidney, posterior urethral valves, etc.) have been reported, as well as vesico-ureteral reflux.

- Facial palsy is significantly associated with renal anomalies, which are usually ipsilateral to the palsy (2)

- Children with CHARGE are at increased risk for urinary tract infection. Evaluation of a patient who has fever without a source should include a urinalysis and urine culture.

Diagnostic tests

1. Baseline renal and bladder ultrasound to evaluate anatomy are warranted, as early identification and treatment may reduce long term morbidity (3,4).

2. Functional studies (such as voiding cystourethrogram) may be indicated as follow-up to anatomic study, or if the patient develops urinary tract infection.

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Medical management and caveats

1. Standard medical therapy is acceptable for treatment of UTIs. Suppressive therapy may be necessary if infections are recurrent.

2. Surgical intervention is indicated for certain abnormalities. Indications for surgery are the same as for children who don't have CHARGE. Caveats for surgery include:

   a. Anesthetic risk is increased in children with airway involvement such as **choanal atresia** or **laryngotracheomalacia** (both common in CHARGE). Children with choanal atresia and complex heart defects have the highest rate of serious complications and/or poor outcome.

   b. Swallowing problems with increased secretions (presumably due to involvement of
   cranial nerves IX and X) may present an additional risk of aspiration.

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THE URINARY TRACT IN CHARGE SYNDROME
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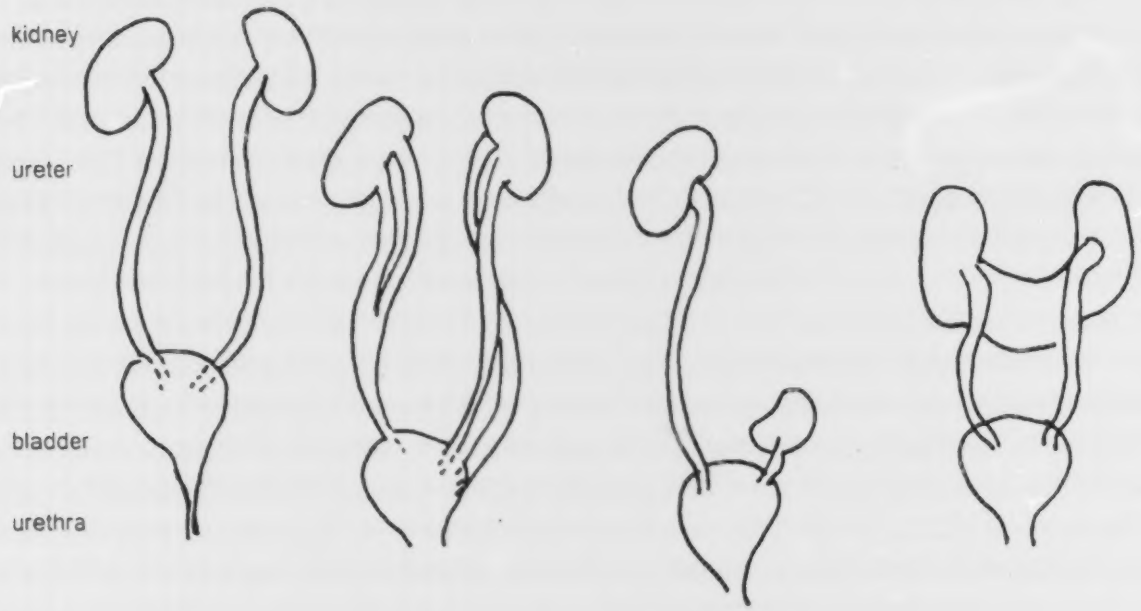
STRUCTURE AND FUNCTION OF THE URINARY TRACT



Normal urinary system | duplicated ureters | pelvic (ectopic) kidney | horseshoe kidney

The normal urinary tract consists of two **kidneys**. Urine is produced in the kidney and is drained by the **renal pelvis**. Urine then flows into the **ureters.** These tubes drain into the **urinary bladder**. When we urinate, the urine travels through another tube called the **urethra**. This tube is very short in females and longer in males (as it has to travel through the length of the penis). Urine normally only flows in one direction.

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URINARY TRACT ABNORMALITIES IN CHARGE

Abnormalities of the urinary tract are seen in 20-40% of children with CHARGE syndrome. They can he of two types, structural or functional.

Structural abnormalities (urinary tract isn't formed properly).

1. Absent or hypoplastic kidney - one kidney is not present or small. If a child is missing both kidneys they cannot survive.

2. Ectopic, or pelvic kidney - kidney is in an abnormal location, much lower than it should be.

3. Horseshoe kidney - the two kidneys are joined together to make one kidney shaped like a horseshoe.

4. Duplications (kidney, renal pelvis and/or urethra can be partially or completely duplicated). Obstruction to urinary flow is frequently seen.

5. Hydronephrosis or enlargement of the renal pelvis - excess fluid into the kidneys. This can be caused by a blockage between the renal pelvis (kidney) and the urethra (which is called a ureteropelvic junction, or UPJ obstruction), or by severe vesicoureteral reflux (see below).

Functional abnormalities (urine doesn't flow properly)

1. Vesicoureteral reflux (hereafter referred to as reflux). When the bladder contracts to assist urination, urine normally flows out the urethra. The bladder wall pinches off the ureters to prevent urine flowing backwards towards the kidneys. If the ureters remain open during bladder contraction, urine can flow back towards the kidney. This is reflux. It can be mild to severe. Reflux can predispose to urinary tract infection (UTI). If severe, it can result in hydronephrosis and direct damage to the kidneys, which can ultimately lead to kidney failure.

2. Bladder residual. Bladder does not empty completely. This can predispose to urinary tract infections.

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DIAGNOSTIC TESTS

1. Renal Ultrasound identifies structural abnormalities.

2. Voiding Cystourethrogram (VCUG) This test is indicated if reflux is suspected. It involves placing a small tube (catheter) through the urethra into the bladder. The bladder is filled with a liquid that can be seen on X-ray (contrast). X-rays are taken while the child urinates. This test is necessary to diagnose reflux or bladder residual.

3. Intravenous Pyelogram (IVP) This is another test primarily used to examine kidney structure. Because it involves the injection of contrast into a vein as well as X-ray exposure, this test has for the most part been replaced by the renal ultrasound. It does have the advantage of requiring intact blood supply to the kidneys and provides information regarding the kidney's ability to make urine (which neither #1 or #2 do). IVP can be used to determine if duplicated kidneys are functioning.

4. Radionucleotide renal scan. This test involves the injection of a radioactive material into a vein that concentrates in the kidney. Indications for the test are similar to #3, but this test gives more information about function and less information about structure than the IVP. The amount of radioactive is small and it is rapidly eliminated in the urine so radioactivity dose is small.

5. Computerized tomography (CT) and Magnetic Resonance Imaging (MRI). Both of these tests provide high-resolution images that allow much better definition of structure than the renal ultrasound. One of these would be indicated if the renal ultrasound was unable to resolve a complex structural abnormality of the urinary tract. The CT scan uses X-rays, but usually does not require sedation as it scans very rapidly. The MRI uses magnetic fields (which do not have harmful effects on body tissues) and has better resolution than CT scan. It can also be used to evaluate blood vessel anatomy (Magnetic Resonance Angiography or MRA). The scan time is longer, so young child frequently require sedation. This is an important issue in CHARGE children due to the increased risk of airway compromise (see section 5f). We recommend full airway precautions, as discussed elsewhere, if sedation is to be used.

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MANAGEMENT OF URINARY TRACT ABNORMALITIES

1. All CHARGE children should have a baseline renal ultrasound done in infancy looking for structural abnormalities.

2. Any child who has fever without an identifiable source on physical examination should have a clean urine specimen obtained looking for UTI. This may involve placing a catheter through the urethra to obtain a specimen or sticking a needle through the abdomen into the bladder (suprapubic or bladder tap). Bagged specimens are inadequate for this purpose. Older children may be able to provide a clean voided specimen. Urine culture must be done to confirm UTI.

3. UTIs can be treated with standard antibiotics. A documented (urine culture positive) UTI is an indication for VCUG.

4. Recurrent UTIs may require use of a daily antibiotic. This is called suppressive or prophylactic therapy. If a VCUG has not been done, it must be done at this point.

5. Surgery is indicated to correct some structural anomalies, and may be indicated in severe cases of reflux. A urologist with pediatric experience should assist with this decision. Follow all surgical and anesthetic precautions as discussed elsewhere.

OUTCOME

Aggressive medical and surgical intervention should be able to prevent or treat most complications of these abnormalities. Rarely, anomalies may be so severe as to inevitably progress to end stage renal disease (ESRD also known as kidney failure) which may necessitate consideration of dialysis or kidney transplant. There is only a single report in the medical literature of this occurring in CHARGE syndrome. (This was in an adult in whom CHARGE syndrome was not recognized until he presented in renal failure. The authors felt that if the syndrome had been recognized early on, diagnostic test could have been done that may have prevented the renal failure).

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MUSCULOSKELETAL ANOMALIES IN CHARGE SYNDROME PHYSICIAN INFORMATION

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TYPE AND FREQUENCY OF MUSCULOSKELETAL ANOMALIES IN CHARGE SYNDROME

- Prevalence of musculoskeletal anomalies is between 30 - 50%. This may increase as more cases of scoliosis are noted in older individuals.
- No consistent pattern of anomalies has been seen, although syndactyly of fingers or toes seems to be more frequent. Some of these patients have an atypical split hand deformity. There is often a specific palmar crease pattern with a so-called "hockey stick" distal palmar crease which is included int eh minor diagnostic criteria.
- Severity of anomalies has ranged from very minor (dermatoglyphic anomalies to moderately severe (congenital hip dysplasia, syndactyly, polydactyly, club foot).
- A single patient is reported with absent muscles of one upper extremity. Muscle abnormalities have otherwise not been reported.
- Hypotonia, particularly of the upper body is frequent. This may be a neurologically-based problem or possibly a misinterpretation of hte presence of ligamentous laxity. There have been no reported cases of a primary myopathy in CHARGE patients. Muscle biopsy would not be indicated, unless a second diagnosis is suspected.
- Scoliosis is frequent, beginning as young as 6-7 years.
- Osteoporosis may occur in adults with untreated hypogonadism.
- Information is derived from literature review of nearly 300 reported patients.

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DIAGNOSTIC TESTS

Careful physical examination of the musculoskeletal system is the only "test" indicated in all patients. It is important to screen for scoliosis beginning at school age.

Other diagnostic tests (X-ray, ultrasound) may be indicated based on physical findings.

X-rays obtained for other reasons (ie. chest x-rays) should be examined carefully for definition of skeletal anatomy.

MEDICAL MANAGEMENT

Treatment is anomaly specific. No differences in therapy are necessary if the patient is diagnosed with CHARGE syndrome. If surgery or sedation is necessary, anesthesia precautions are as discussed elsewhere.

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DEVELOPMENTAL EFFECTS OF VISION LOSS (also see Development sections)

Infants with decreased vision will have delayed motor milestones. This is especially true for children with CHARGE, who often also have hearing loss, vestibular (balance) abnormalities and serious medical problems requiring multiple hospitalizations and surgeries.

Communication can be complicated by vision problems in children with CHARGE. Because of the hearing loss, sign language, speech reading and other visual communication is often used. Decreased visual acuity can make this more of a challenge.

MEDICAL TREATMENT OPTIONS

There is no medical or surgical treatment for coloboma

Retinal detachment can often be treated surgically if it is correctly diagnosed shortly after it occurs. Suspected retinal detachment should be regarded as a medical emergency.

Amblyopia may be treated with patching, strabismus may be treated with surgery.

Refractive errors that reduce visual acuity can often be helped with glasses.

NONMEDICAL MANAGEMENT

Photophobia can be helped by using tinted glasses, indoors as well as outdoors.

It is important to know the extent of your child's visual field. If there is very little vision in the upper portion of the visual field, sign language and objects the child may want to see must be placed in the lower half of the visual field (in the lap). Many children compensate for small visual fields by adopting a certain head position or body position.

Education

If your child has both hearing loss and vision loss (even if she or he is not "deaf" or "blinid"), the educational team for your child should include a specialist in deaf-blind children. Such specialists exist in every state in the U.S. Input from such an expert is important even if the hearing loss or vision loss is not "complete."

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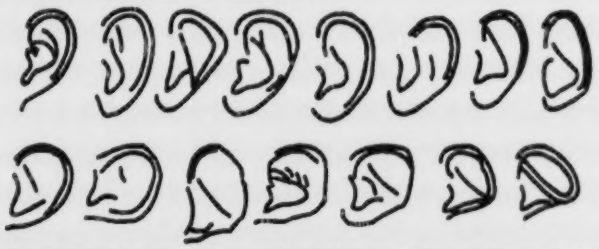
THE EARS IN CHARGE – for the Physician

Sandra L.H. Davenport M.D Sensory Genetics/Neuro-Development, Bloomington MN
Udayan K.Shah M.D. Pediatric Otolaryngology, CHOP, Philadelphia PA

I. Outer ears & canals

A. Distinctive shape of CHARGE Ears[1] (see diagram)

1. Ear shape can be so distinctive as to tip off the examiner to a diagnosis of CHARGE:
   a) Helical fold: thin, "clipped off" along the inferior edge or absent
   b) Antihelix: may extend out to the helical rim rather connecting smoothly to the antitragus
   c) Tragus: Usually intact
   d) Antitragus: Usually present or thinned but may not connect to antihelix to form the posterior rim of the concha
   e) Concha: Takes on a more triangular appearance as the antihelix extends farther out
   f) Lobules are frequently small or absent.



2. Lop ears are fairly common but in and of themselves do not constitute "CHARGE ears" When the ear is put into the normal position, look for the distinctive CHARGE features.

B. Floppy ears due to soft cartilage

Typically, the pinnae flop forward or can be bent forward too easily even after the newborn period. The reason for the apparently abnormal cartilage is not known. It does firm up somewhat over months or years but even adults may have somewhat soft pinnae.

C. Reconstruction

Taping back the ears during the few months of life can partially correct a lop-ear deformity.[3] Though otoplasty can serve a cosmetic purpose in older children, it has been done primarily to provide better support for behind-the-ear hearing aids and eye glasses. However, the weight of the aids may push the ear back into the preoperative position. Additional means of securing the aids should be used until the cartilage is firm enough to support the weight.

D. Canals & Tympanic Membranes

Significantly stenotic canals are rare in CHARGE. TMs appear normal unless change in position or shape reflects ossicular changes or severe Eustachian tube trouble.

II. Middle ears

A. Ossicular malformations

The possibility of ossicular malformations is frequently overlooked when chronic serous otitis media is present. Therefore, audiometry after PE tube placement is important. CT scan of the middle and inner ear is important in all cases where any hearing loss is present in order to rule out malformations.

B. Chronic serous otitis media

CSOM is common with or without accompanying cleft palate. Because of the detrimental effect any additional hearing loss has on development, *PE tube placement should not be delayed.*

III. Inner ears

A. Mondini malformations

Significant malformations are common but much underappreciated. Early confirmation by CT scan is helpful for medical/habilitation prognosis and management.

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B. Cochlear involvement

Hearing losses range from none to profound. Sensorineural loss is usually stable and aidable so management is the same as in other conditions. Cochlear implants have been done successfully and can provide at least environmental information for deafblind children. Keeping hearing aids on and working is very difficult. The reasons include not only the floppy cartilage mentioned above but also the frequent presence (90%) of impaired swallowing so that copious oronasal secretions run down the face of the supine child into the ear. The secretions may loosen the ear mold or block the air hole. In addition, the secretions may keep the canal moist setting up the conditions for chronic otitis externa.

C. Vestibular involvement

Congenital loss of vestibular function is probably the single most important factor in delayed gross motor milestones so early imaging of the inner ear is indicated.[5] This will decrease the need for extensive neurologic testing and falsely low estimations of intelligence IF the implications are communicated to the primary physician and neurologist.

Few tests other than tomography are available to confirm vestibular involvement in early childhood. Caloric testing may help though many children have PE tubes or TM perforations by the time this question comes up. Placing the baby in the mother's lap in a rotatory chair and comparing their eye movements is possible only if the child does not have a significant coloboma affecting the macula (central vision). Posturography is usually not possible until middle childhood because walking is often delayed to 4-5 years, visual fixation may be impaired by the colobomas, and the child will not cooporate if he/she does not clearly understand what to do.

D. Retrocochlear involvement

Very little firm data is known though some suggestion of central hearing loss has been made.[2] Since surrounding cranial nerves, such as VII, IX, X are clearly involved, it would make sense that the VIII nerve nuclei could be involved also. We know of no postmortem confirmation of this.

IV. CAVEATS: What make CHARGE different from other multiple malformation syndromes?

Multiple cranial nerve involvements produce many ENT concerns: olfactory, facial, glossopharygeal and vagus nerves may be involved. Choanal stenosis/atresia, cleft lip/palate and TE fistulas may also be present. The ENT consultant is an early and very important member of the team.

Dual sensory loss is present in over 3/4 of cases. Therefore, typical hearing habilitation measures may be inappropriate since they rely on good vision. INTELLIGENCE IS ROUTINELY UNDERESTIMATED in these children. Early identification of hearing and vision loss is often made without early referral to the appropriate education team, which should include educators familiar with dual sensory losses. This referral should be made as ASAP since communication is challenging to establish and yet is the key to assessing intelligence and optimizing quality of life.

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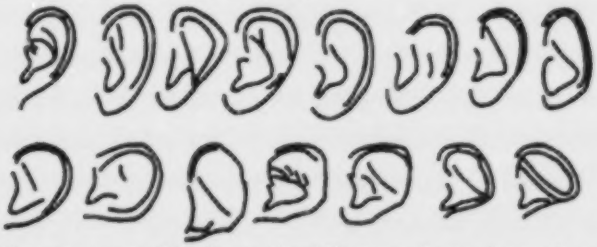

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THE EARS AND HEARING IN CHARGE SYNDROME – PARENT INFORMATION

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Abnormalities of the ears, ear infections, and hearing loss are very common features in CHARGE. Together these problems can affect health and the ability to speak and learn. As with other aspects of CHARGE, not every individual has every problem. The number of problems each individual has and the severity of each problem varies greatly – within individuals and across individuals. As a result, each individual's set of abilities and disabilities is unique.

HEALTH CARE PROVIDERS FOR EARS AND HEARING

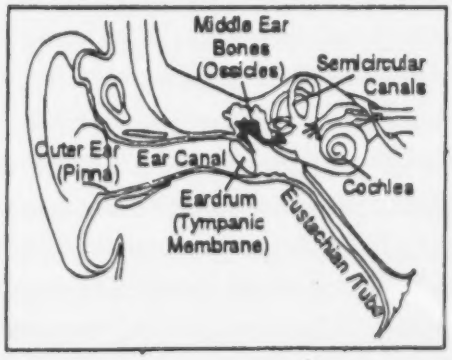
Medical care for ears and hearing in CHARGE is best provided by a physician who specializes in ears: an otologist, otolaryngologist, or ear-nose-and throat (ENT) physician. Though ear problems in childhood are often diagnosed and treated by the primary medical care provider (family physician or pediatrician), the problems in CHARGE are so complex that they need to be diagnosed by an otolaryngologist who makes an otologic (ear) diagnosis and recommends a plan of treatment to the primary care provider.

Hearing is assessed by an audiologist. The results of the hearing assessment are used in the medical diagnosis of the hearing loss, in determining the success of medical/surgical treatment, and in developing an aural habilitation plan. This is a plan for overcoming hearing loss that cannot be corrected medically or surgically. It can include selection and fitting of hearing aids, speech-language evaluation/therapy, teaching of sign language, and placement in educational programs.

It is very difficult to assess hearing accurately on persons with CHARGE – especially infants and young children. In some cases, detailed hearing evaluation many require many test sessions. Until hearing is assessed with confidence, it is often difficult to establish the otologic diagnosis, to select hearing aids, or to make optimal educational recommendations.

Children with CHARGE need care from otolaryngologists and audiologists who specialize in the disorders of children and especially children with unusual problems. Many audiologic test facilities are not equipped to perform the types of tests needed for children with CHARGE. Selection of the appropriate otolaryngologist and audiologist is crucial to the accurate diagnosis and successful treatment of ear and hearing problems in CHARGE.

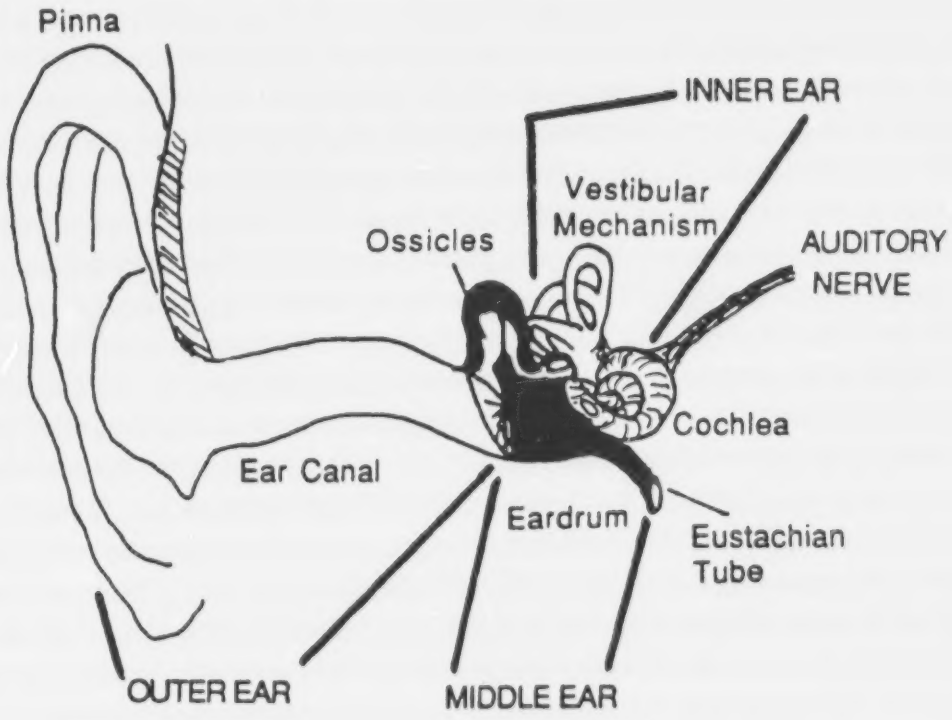
EAR AND HEARING ABNORMALITIES IN CHARGE SYNDROME



The structures of the ear shown in in the diagram and described below.

FIGURE : STRUCTURES OF THE AUDITORY SYSTEM

OUTER & MIDDLE EARS
(Conductive Hearing Loss)



INNER EAR & AUDITORY NERVE
(Sensorineural Hearing Loss)

OUTER AND MIDDLE EARS

The outer and middle ears conduct sound to the inner ear. Hearing loss due to problems in the outer or middle ears is called "CONDUCTIVE HEARING LOSS." Conductive hearing loss only makes the sound that reaches the inner ear softer. Amplifying the sound overcomes the hearing loss by restoring loudness and clarity. Conductive loss is common in CHARGE, but it is usually only part of the total hearing loss.

Outer Ear (Pinna)

Pinnas can be slightly or significantly deformed. Unless the ear canal is blocked by tissue, deformities of the pinna have almost no effect on hearing. However, if a hearing aid needs to be worn, the pinna may be too soft or not large enough to accommodate a behind-the-ear hearing aid that is commonly fitted on a child. Special devices may be required to keep the hearing aid in place. In some cases, pinna shape can be improved with surgery.

Ear Canal (External Auditory Canal)

The ear canals in CHARGE can be narrow (stenotic). This does not usually affect hearing, but it may make it difficult to examine the eardrum and middle ear, to clean wax (cerumen) from the ear, and to fit a hearing aid earmold in the ear canal. If the ear canal is not present (atresia), it causes a large conductive hearing loss. In some cases, surgical construction of an ear canal may be attempted.

Middle Ear

The middle ear is an air-filled space behind the eardrum (tympanic membrane). Sound moves the eardrum and the three tiny bones (ossicles) which transfer the motion to the inner ear. In CHARGE, it is common to have malformed ossicles that cause significant conductive hearing loss – especially for low pitch or low frequency sounds. Usually, no attempt will be made to correct this problem with surgery.

At the bottom of the middle ear cavity is the Eustachian tube that connects the middle-ear cavity to the back of throat. When the Eustachian tube functions normally, it permits fresh air to enter into the middle-ear cavity and it balances the pressure on either side of the eardrum so that it can work optimally. If the Eustachian tube does not open regularly, air in the middle-ear cavity is resorbed into tissue and a vacuum is formed in the middle ear that causes the eardrum to be retracted. This causes a slight to mild low-frequency conductive hearing loss. If the Eustachian tube ventilates the middle ear, the loss vanishes. If the Eustachian tube does not open, fluid may collect in the middle ear (otitis media with effusion) and cause conductive hearing loss that is painless. If the fluid becomes infected, the retracted eardrum becomes a bulging eardrum that is reddened and painful (acute otitis media). In these cases, immediate medical treatment – usually with antibiotics – is needed to prevent rupture of the eardrum. Successful treatment of the infection eliminates pain but does not necessarily eliminate fluid behind the eardrum or the temporary hearing loss caused by the fluid.

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Pressure-equalization tubes (PE tubes) are often inserted surgically in the eardrums to temporarily overcome the problems brought on by poor Eustachian tube function. PE tubes are shaped like sewing bobbins and are a little bigger than the tip of a ball-point pen. They allow air to flow into the middle ear when the Eustachian tube will not open. They typically remain in place from several months to a year before they fall out. Many children with CHARGE require several sets of PE tubes. A marked improvement in hearing is often noted after the insertion of PE tubes.

Inner Ear (Cochlea)

Abnormalities of the inner ear or cochlea are a major cause of permanent hearing loss in CHARGE. The cochlea is named for its shape – snail shell in Latin. It is a normally fluid-filled structure that is smaller than the tip of the little finger, and it is embedded in the hardest bone in the body. It is the sensory end organ for hearing. It changes motion into neural impulses that can be processed as sound by the brain. Damage to the inner ear and the nerves that carry the messages to the brain is called "SENSORINEURAL HEARING LOSS."

When the shape of the cochlea or the vestibular mechanism (which is joined to the cochlea) is abnormal, it may be detected using radiographic procedures (CT or MRI). However, there may be sensorineural hearing loss even though the shape of the cochlea is normal.

Sensorineural hearing loss creates several problems. Sounds may not be heard unless they are amplified. However, when they are amplified, they may be distorted in a way that they cannot be understood. For example, speech may always sound muffled or garbled even with the best hearing aid. Lastly, there may be "loudness recruitment" in which amplified sounds become intolerably loud. In fitting hearing aids on individuals with sensorineural hearing loss, it is necessary to amplify enough so that speech may be heard but not too much so that speech and other sounds become intolerable. If either goal is not met, the hearing aid may be rejected.

Neural Hearing Loss and Central Auditory Processing

Abnormalities of the nerves leading from the cochlea to the brainstem and from the brainstem to the brain do not cause hearing loss in the conventional sense. If a person had this type of loss, they may have normal hearing thresholds but a poor ability to understand speech under any conditions and especially in background noise. These are problems that receive audiologic and educational treatment rather than medical or surgical treatment.

Mixed Hearing Loss

When both conductive and sensorineural hearing loss is present, the loss is called "MIXED HEARING LOSS." This is the most common type of hearing loss in CHARGE. It is usually composed of permanent conductive loss due to malformation of the ossicles, fluctuating conductive loss due to the intermittent presence of fluid in the middle ear, and sensorineural hearing loss that usually was present at birth. In general, the conductive losses are greatest for the low frequencies and the sensorineural losses are greatest for the high frequencies. Often there is marked asymmetry in hearing between the two ears.

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Progressive Hearing Loss

Progressive hearing loss (conductive and/or sensorineural) is an uncommon finding n CHARGE, but has been reported. However, there is probably insufficient audiological documentation over time on a group of children to reach a good conclusion. In cases where the loss has progressed, it has usually been a small increase superimposed on a large existing loss.

As the child matures and communication improves, behavioral hearing thresholds often improve slightly because of the child's improved ability to concentrate or to perform the listening task, rather than a change in hearing. Thresholds obtained with ABR (BAER) tests should not improve markedly with maturation.

HEARING ASSESSMENT

Pure-tone Audiogram

The most important information to be obtained in an audiologic evaluation is the determination of the thresholds of hearing for specific frequencies for the two ears measured independently. The information is recorded on a form called the "AUDIOGRAM." An example of an audiogram is shown in Figure 1 (next page). The thresholds of hearing can be measured in behavioral tests in which the individual is required to give an active response to sound or using an auditory brainstem response (ABR) test which requires that the person be asleep or sedated. Usually, attempts are made to obtain responses to pure-tone signals over a range of test frequencies that is important for understanding speech (500, 1000, 2000, and sometimes 4000 Hz). This information is very important to the otologic diagnosis and the decisions related to hearing aids. Other tests may be done, but this is the most important. The audiologic evaluation process should be repeated until there is confidence in the pure-tone hearing thresholds that have been measured.

Tympanometry

This is a test of the mobility of the eardrum and middle ear. It requires only that a child sit still for a short period of time, and it does an excellent job of detecting the presence of Eustachian tube dysfunction and fluid in the middle ear. This test is used regularly in the management of middle-ear disease – however, it is not a test of hearing. "TYMPANOGRAMS" give information about the middle ear and not the overall ability to hear.

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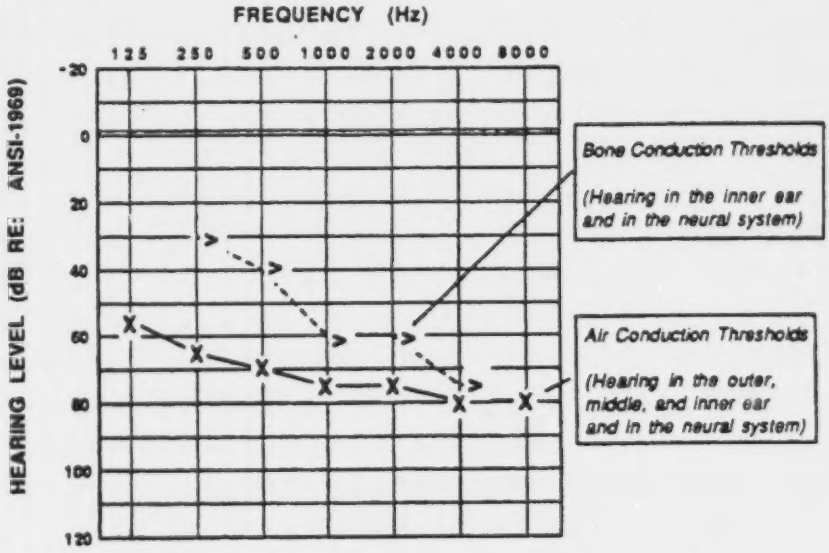
FIGURE : AUDIOGRAM

The audiogram is a graph showing hearing thresholds measured in dB Hearing Level (HL) --on the vertical axis -- for signal frequencies from 125 Hz (low pitch) to 8000 Hz (high pitch).

-AIR-CONDUCTION THRESHOLDS (OVERALL HEARING LOSS)
When test is performed using earphones, the results are called air-conduction thresholds. The air-conduction thresholds indicate the total hearing loss at each frequency due to problems in the outer, middle, and inner ear as well as due to auditory nerve damage. The symbols used to record air-conduction thresholds O and Δ for the right ear and X and □ for the left ear.

-BONE-CONDUCTION THRESHOLDS (HEARING LOSS DUE TO INNER EAR AND NERVE)
Bone-conduction thresholds are measured with a vibrator placed behind the ear. The vibrator bypasses the outer and middle ears and stimulates the inner ear directly. If the bone-conduction thresholds are not normal there is sensorineural hearing loss which usually reduces the ability to understand speech even when it is heard. The symbols for bone conduction thresholds are < and [for the right ear and > and] for the left ear.

Mild hearing loss is greater than 25 dB HL; profound hearing loss is greater than 90 dB HL. The most important frequencies for understanding speech are 500, 1000, and 2000 Hz.



The audiogram above shows an audiogram that is common with CHARGE syndrome. The overall loss is a moderately severe to severe mixed hearing hearing loss in the left ear -- shown with the X's: 55 dB HL at 125 Hz sloping gradually to 80 dB HL at 8000 Hz. The loss is a mixed loss because it has sensorineural and conductive components:

-There is a sensorineural (inner ear and/or nerve) loss -- shown with the >'s. It is mild at 250 Hz (30 dB HL) and severe at 4000 Hz (75 dB HL). This shows that damage to the inner ear is greatest in the higher frequency regions.

-There is a conductive (outer and/or middle ear) loss – represented as the gap between the > and X symbols: 35 dB at 250 Hz and 5 dB at 4000 Hz. The conductive loss is mild in the low frequencies and very small in the high frequencies.

AURAL HABILITATION

Case Manager for Aural Habilitation

The aural habilitation program needs to be managed by a professional who is in constant contact with the child with CHARGE. This may be an audiologist associated with the treating otolaryngologist or a professional associated with the educational system (audiologist, speech-language pathologist, special educator, teacher, etc.). If there is a significant hearing loss, the educational progress needs to be monitored closely and the amplification system needs to be checked daily.

Amplification

When the hearing has been measured adequately, hearing aids need to be selected and fitted by the audiologist. It is important to remember that the sole purpose of hearing aids is to improve hearing, and that the aids have little value if they do not achieve that goal. In children with CHARGE, this is very difficult to achieve but it is the goal. The hearing aids must stay on the head, the ear molds must not hurt or irritate the ear canals, there must be adequate amplification but not overpower the child, and the frequencies amplified must improve the ability to understand speech.

In addition to hearing aids, there are amplification devices called assistive listening devices or auditory trainers. These devices have a microphone that is located close to the talker so that the listener has a better chance to hear. Assistive listening devices are valuable when there is background noise – especially in classrooms.

The type(s) of hearing aids selected will depend on a large number of factors. In-the-ear (ITE) aids are usually not appropriate for children. Behind-the-ear (BTE) aids are often chosen because they have power and flexibility. Body-worn aids are also considered if the hearing loss is very large. Cochlear implants are surgically implanted devices that bypass the cochlea and electrically stimulate the nerves directly; they are only considered when the losses are in the severe to profound category. If there is any measurable benefit from hearing aids, it is unlikely that cochlear implant surgery would be performed. There are strict FDA guidelines for who may be considered for implantation. Very few children with CHARGE have received cochlear implants.

DEVELOPMENT AND EDUCATION

For many children with CHARGE, the first years of life are spent treating life-threatening problems. After this period the parents have the chance to focus on development and education. It is important to note that most children with CHARGE pass through critical developmental periods for the development of speech and language in a state of sensory deprivation. Usually there have been significant hearing and vision problems regardless of attempts to overcome them. As a result the child with CHARGE begins the educational process at a disadvantage. In whatever educational a child with CHARGE is placed, there needs to be a coordinated effort of all the professionals who have knowledge of the child's abilities and disabilities to ensure success.

CHOANAL ATRESIA IN CHARGE SYNDROME - for Physicians

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ANOMALIES SEEN IN CHARGE

A little more than 50% of children with CHARGE have some form of choanal atresia. The range is complete - from bilateral bony choanal atresia to unilateral choanal stenosis. About half of the patients with choanal atresia have bilateral choanal atresia. In the general population, the incidence of choanal atresia is approximately 1 in 5000-7000 live births. A large percentage of these probably represent children with CHARGE.

DIAGNOSTIC TESTS

Physical Exam

Once the diagnosis of choanal atresia has been made, it can be confirmed initially on physical exam by failure to pass a # 6 to 8 French plastic catheter through the nares into the pharynx. (a typical solid feeling will be encountered at the level of the posterior choanal approx. 3-3.5 cm from the alar rim). Passage of soft metal probes has also been proposed.

Imaging Studies

Plain films and tomograms of the skull with radiopaque dye instilled into the nasal cavities can confirm choanal atresia. An axial noncontrast high resolution CT scan with thin sections (2-5mm) has become the single radiographic study of choice. The CT scan has proven invaluable in the accurate assessment of both the normal and abnormal anatomy of the nasal cavity, posterior nasal choanae and nasopharynx. The knowledge obtained from the CT scan is valuable in the preoperative planning of the method and design of the repair.

When planning a CT scan or MRI in a child with suspected CHARGE, consult with otolaryngology: often a few more cuts can yield important information about the inner ear abnormalities seen in CHARGE.

CONSEQUENCES OF CHOANAL ATRESIA IN CHARGE

Bilateral choanal atresia

Bilateral choanal atresia causes complete nasal obstruction - immediate respiratory distress and even potential death due to asphyxia (because newborns are obligate nose breathers until approximately 4 to 6 weeks at which time mouth breathing is learned). The respiratory obstruction is cyclic - as the child falls asleep the mouth closes and a progressive obstruction starting with stridor followed by increased respiratory effort and cyanosis. Either the observer opens the child's mouth or the child cries and the obstruction is cleared.

Initial feeding is often the alerting event - as the child starts with inability to eat and breath at the same time, there is a progressive obstruction of the airway and subsequent cyanosis and choking due to aspiration of the milk. This can mimic a tracheoesophageal fistula (TEF). TEF and/or esophageal atresia is common in CHARGE.

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Unilateral choanal atresia

Unilateral choanal atresia rarely causes any acute respiratory distress. The most common finding is a unilateral mucoid discharge. Unilateral choanal atresia does not require any immediate surgical attention, but may eventually require treatment because children with CHARGE have numerous other potential respiratory complications which can be exacerbated by UL choanal atresia.

MEDICAL MANAGEMENT

An oral airway of some sort must be implemented very early on in the treatment of newborns with choanal atresia. A typical anesthesia oral airway is often sufficient, however if not then orogastric tube may be considered. A large nipple can be modified by having its end cut off and then ties are attached to the nipple and placed around the occiput. This type of airway is called a McGovern nipple and provides an airway through which the baby can breathe. A very small feeding tube can then be passed either through another hole in the nipple or along side the nipple for gavage feeding. This is the preferred method of establishing an oral airway.

Tracheotomy and caveats for CHARGE

This is a controversial issue and many physicians conclude that there is never a need for this drastic a step in the initial management of infants with choanal atresia. Patients with CHARGE have a high propensity of airway instability. These children's early repair of their choanal atresia is rarely successful, primarily due to their abnormal anatomy of their nasopharynx and upper aerodigestive tract. Many children with CHARGE have at least one cardiopulmonary arrest prior to their definitive procedure. Therefore, some investigators have proposed that children who have CHARGE should have early tracheotomy to protect their brain from anoxic injury and delay the repair of their choanal atresia until they are at lease 2 years of age. This is still a very controversial subject.

Feeding

Gavage feeding is usually required until the child has learned to mouth breath. Then once the child learns the mouth breath, oral feeding can be attempted. A majority of children with CHARGE have significant feeding problems, possibly related to abnormalities of cranial nerves IX an X. Many of these children require gastrostomy feeding for a number of years. See sections on Swallowing and Growth.

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CHOANAL ATRESIA for Parents

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NORMAL STRUCTURE AND FUNCTION

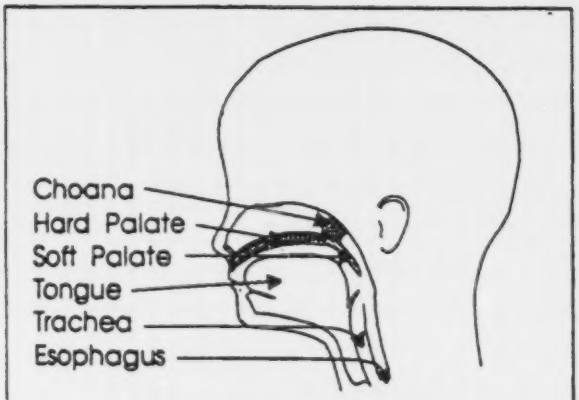

Fig. 1 Normal anatomy showing cut-away view of funnel-shaped open choana at rear of right nasal passage.

The choanae are funnel-shaped openings at the back of the nasal passages which connect the nose with the throat. They are located just above the roof of the mouth, where the hard and soft palate meet (figure 1).

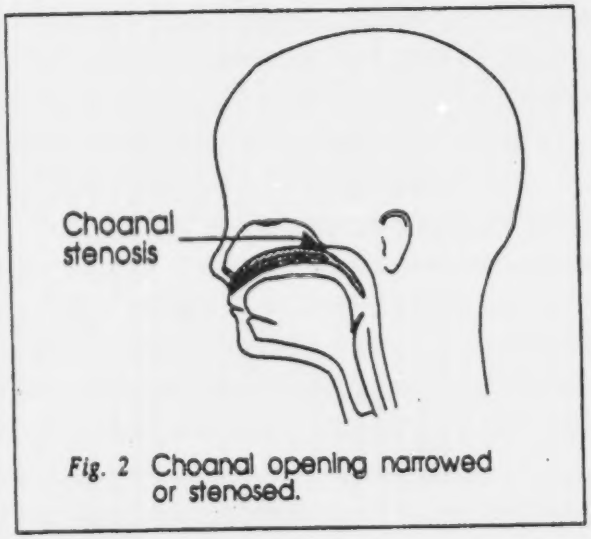

Fig. 2 Choanal opening narrowed or stenosed.

In normal fetal development, the choanae open when a membrane breaks down at about the seventh week of pregnancy. If this membrane does not break down, choanal atresia results. If the blockage is only partial, the term choanal stenosis (meaning tight or narrow) is used (figure 2). Choanal atresia is total blockage between the nose and the throat (figure 3).

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The choanae are critical in the newborn because newborn babies are obligate nose-breathers. If both nasal passages are totally closed off (bilateral choanal atresia), the newborn child almost always has trouble breathing. Babies cannot instinctively switch from nose breathing to mouth breathing. This can lead to lack or air (asphyxia) with lack of oxygen (anoxia) which can result in respiratory distress, brain damage, or death if not recognized and treated quickly.

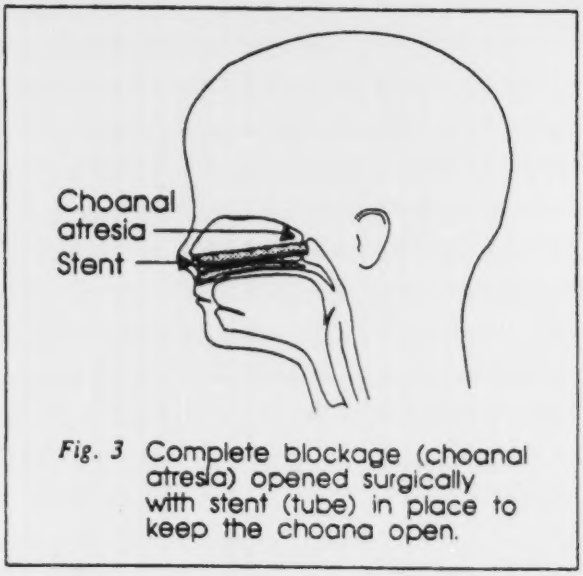

Fig. 3 Complete blockage (choanal atresia) opened surgically with stent (tube) in place to keep the choana open.

Fig. 3
Complete blockage (choanal atresia) opened surgically with stent (tube) in place to keep the choanal open.

About half of all children with CHARGE have some form of choanal atresia. It is very rare in other conditions, and therefore very helpful in making a diagnosis of CHARGE. Children with CHARGE can have choanal atresia (blockage) or choanal stenosis (narrowing). It can affect both sides (bilateral, BL) or only one side (unilateral, UL). Choanal atresia can be bony (bone or cartilage blocking the passageway) or membranous (soft tissue membrane blocking the passageway).

Choanal atresia can be present even if there is a cleft palate. It may be more difficult to diagnose, as air can move through the cleft. Unilateral choanal atresia and choanal stenosis can be very difficult to recognize.

DIAGNOSIS OF CHOANAL ATRESIA

Polyhydramnios (excess amniotic fluid) during pregnancy is often the first sign of choanal atresia. Normally, the fetus breathes amniotic fluid during pregnancy. If the nose is blocked, and the fetus can't "nose breathe," the amniotic fluid is not "recycled" by fetal breathing motions and excess fluid may build up in the uterus.

A tube should be passed through each nostril and nasal passage of any infant who has unexplained breathing difficulties in the first month of life. This will establish if the nasal passages are open (patent). X-rays can be performed using dye in the nasal passages to verify they are not open. Sometimes other imaging (MRI, CT) is used to confirm the diagnosis and determine the extent of the problem.

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TREATMENT

Bilateral choanal atresia

Bilateral choanal atresia is life threatening in the newborn period, and treatment is urgent. Emergency treatment usually consists of placing a plastic airway (tube) into the mouth to keep the mouth open. This allows the baby to mouth-breathe. Other newborns may require intubation: passing a breathing tube through the mouth and down into the windpipe (trachea) so oxygen goes directly into the lungs. Occasionally, it is necessary to do a tracheotomy: surgically putting a breathing tube directly into the trachea through a hole in the lower front of the neck.

The above procedures are all temporary. Soon after, surgery will be performed to open the bone or membrane covering the nasal passage. A stent (plastic tube) is placed in each nasal passage to keep it open and guarantee adequate air entry (figure 3). The stents may have to stay in for a few weeks or even months. Children with CHARGE are more likely to have complications following choanal atresia repair than other children with choanal atresia. A few require repeat surgery later because the choanae close up again when the stents are removed.

Unilateral choanal atresia and/or choanal stenosis

Lesser problems usually occur when one nasal passage is totally obstructed (UL atresia) or when one or both nasal passages are narrowed (stenosis). Air can pass in varying amounts through the nose into the lungs. The child with UL atresia or stenosis often does not have obvious respiratory symptoms, but he or she may have a constant runny nose on the side that is blocked or narrow. Recognizing choanal stenosis or unilateral atresia can be important in the treatment of these respiratory complications.

OUTCOME

A major long-term complication of choanal atresia or stenosis is recurrent ear infections, which can lead to a conductive hearing loss. Because children with CHARGE also have a high risk for sensorineural hearing loss, these ear infections should be treated aggressively.

Children with CHARGE may require more than one surgery before the choanal atresia is permanently repaired. Often, difficult decisions must be made about surgery - should the heart be fixed first or the choanal atresia? Is the baby strong enough for surgery, given the heart problems, etc. Any newborn with CHARGE should have the choanae evaluated before any surgery is done because of possible complications. Choanal atresia and the resultant lack of oxygen increases the risk for mental retardation due to brain damage.

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OROFACIAL CLEFTING IN CHARGE SYNDROME FOR THE PHYSICIAN

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CRANIOFACIAL ANOMALIES SEEN IN CHARGE

Orofacial clefting occurs in about 20% of children with CHARGE syndrome. These children may have cleft lip with or without cleft palate or isolated cleft palate, especially sub-mucous cleft palate.

DIAGNOSTIC CONSIDERATIONS:

In general, patients with CHARGE are more likely to have cleft lip, while those with velo-cardio-facial syndrome (VCFS) are more likely to have a cleft palate. When orofacial clefting is present in patients with CHARGE, the choanae are usually patent, so this finding (clefting) may substitute for choanal atresia in the diagnostic criteria, particularly if the remaining findings are otherwise characteristic of this condition.

Other anomalies common in CHARGE which may affect treatment and/or management of orofacial clefts:

- TE fistula or esophageal atresia
- Cranial nerve IX/X palsies
  - Laryngotrachomalacia
  - Velopharyngeal incompetency
  - Reflux
- Facial palsy, unilateral or bilateral
- Heart defects
- Ossicular malformations with or without Mondini anomaly
- Choanal atresia or stenosis

DIAGNOSTIC TESTS:

While cleft lip is obvious to the casual observer, the diagnosis of submucous cleft palate in CHARGE can sometimes be delayed. Many cases have been confirmed only when tonsils or adenoids are removed. **An evaluation looking for cranial nerve IX/X involvement is crucial prior to beginning a feeding program in order to avoid multiple aspiration pneumonias.**

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CONSEQUENCES OF OROFACIAL CLEFTING IN CHARGE:

FEEDING:
Orofacial clefting can interfere with feeding in any individual. Children with CHARGE often have additonal feeding problems due to neurologially-based velopharyngeal incompetency and/or reflux. Cleft lip or cleft palate can exacerbate this situation. Children with CHARGE (with or without orofacial clefting) often require g-tube feeding for significantly longer than children with other clefting syndromes.

EAR INFECTIONS/HEARING LOSS:
Children with facial clefts are prone to ear infections and possible hearing loss. Children with CHARGE typically have more ear infections and require PE tubes more often and for a longer period of time than other children with clefts. Children with CHARGE often have sensorineural hearing loss and/or conductive hearing loss due to malformed ossicles. Close follow-up by ENT and audiology is critical to maximizing the hearing in these children.

SPEECH:
An undiagnosed submucous cleft palate can interfere with speech development, already complicated in these children due to hearing loss and facial palsy.

MEDICAL MANAGEMENT AND CAVEATS
Team management approach: It is the recommendation of the American Society of Maxillofacial Surgery as well as the American Society of Plastic and Reconstructive Surgeons that management of the patient with facial clefting be provided by an interdisciplinary team of specialists offering a coordinated and consistent philosophy and a continuum of care. The cleft team may include a plastic surgeon, oral surgeon, orthodontist, otolaryngologist, ophthalmologist, social worker, nurse, audiologist, speech pathologist, and geneticist. When following a child with CHARGE, it is important to communicate with other specialists who are following the child. The feeding team members should understand the neurologic basis for incoordinated swallow and reflux. The members of the team and the approach to the child will vary from one institution to another. The specific managment plan for orofacial clefting in a child with CHARGE will vary depending on the specific problems of that child as well as the protocols for a particular institution.

Early simple surgical repair maximizes optimal speech, aids in feeding, assists in reducing middle ear infections, and re-establishes normal separation of the oral and nasal cavity while minimizing growth disturbances of the upper jaw.

Post-surgical complications such as infection, dehiscence, oro-nasal fistula, and malocclusion may be more frequent in children with CHARGE. Therefore, a 23-hour stay planned for children with isolated clefts may need to be extended to an inpatient stay for children with CHARGE. **Consider insertion of PE tubes and the taking of dental impressions at the time of surgery if indicated.**

Bilateral cleft lip and palate is rare in CHARGE syndrome. Although late treatable complications such as dental malocclusion, velopharyngeal incompetence, oro-nasal fistulae, and distortion of normal anatomy can often occur, initial planning during the first twelve months of life mirrors that of unilateral cleft lip and palate.

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SPECIAL CONSIDERATIONS IN TREATMENT OF FACIAL CLEFTS IN CHARGE:

1) Heart defects (present in 2/3 of children with CHARGE) may be complex. These may effect timing of surgery and overall stability of the patient.

2) Possible unexpected reaction to anesthesia. Some children with CHARGE are resistant to sedation, while others are slow to recover from anesthesia. Because of risks of anesthesia, it may be appropriate to combine surgical procedures in these children.

3) Laryngomalacia or tracheomalacia is common in CHARGE and can result in surgical or anesthetic complications.

4) Cranial nerve palsies complicate post-operative feeding in a major way. Most importantly, apparent abnormalities of cranial nerves IX and X cause secretions and food to pool in the pharynx. Esophageal peristalsis is uncoordinated. Aspiration and reflux are common. This improves in weeks, months or years, with a few children having permanent problems. In addition, facial palsy is usually unilateral and may affect lip closure and may compromise ultimate cosmetic outcome of cleft lip.

NON-MEDICAL MANAGEMENT

If a patient has CHARGE, orofacial clefting is likely to be only one of a number of serious medical problems for that child. The cleft may or may not be a high-level concern to the parents. It is critical to talk to the parents about the whole child, and not just about the cleft. Parents are often especially concerned with how the cleft correction fits into other surgical schedules.

The orofacial team which includes ENT, audiology, and feeding specialists can be a great help to parents (even if the child does not have a cleft) because they have experience with feeding difficulties, PE tubes, and ear infections, all of which are common in CHARGE. Communication with other specialists following the child is critical to effective care of these children. These children often have multiple medical appointments every month or even every week, so coordination of appointments is appreciated by the parents.

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CLEFT LIP AND PALATE (CL/P) IN CHARGE - PARENT INFORMATION

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NORMAL STRUCTURE AND FUNCTION:
You know what the lips are! The **palate** is the roof of the mouth: the bony and muscular structure between the oral (mouth) and nasal (nose) cavities.

Embryology:

The lips and palate develop between 5 and 7 weeks of gestation (pregnancy). They result from the growth, merging, and fusion of five "processes:" two maxillary (upper jaw) processes, two mandibular (lower jaw) processes, and one frontonasal (nose) process (see diagram). The lower lip is formed when the mandibular processes unite. The upper lip is formed from the two maxillary processes and the frontonasal process. They come together (two from the side, and one from above) to form the upper lip. The "cupids bow" in the center of the upper lip shows the borders where the processes meet.

The primary (hard) palate, the nasal cavity and the choanae (passages from the back of he nose to the throat) result from merging of the medial nasal processes at 5 weeks. At 6 weeks, the secondary (soft) palate processes develop and fuse. This creates the soft palate, the nasal cavity and the choanae. The lips, palate and nasal cavity should be complete by about 7 weeks gestation. If any of these processes fail to fuse or merge, a gap, or cleft, results.

FREQUENCY OF OROFACIAL CLEFTING IN CHARGE:

Orofacial clefting (cleft lip or cleft palate) occurs in about 20% of children with CHARGE syndrome. Submucous cleft palate is often not diagnosed in the newborn period, so the frequency of clefting may be even higher. It is possible to have both choanal atresia and cleft palate.

Diagnosis of Clefts in CHARGE
Cleft lip is obvious at birth. The newborn exam performed in the delivery room usually includes an evaluation of the palate. A cleft of the hard palate should be apparent as part of this exam. A submucous cleft is more difficult to diagnose and is sometimes not recognized until much later.

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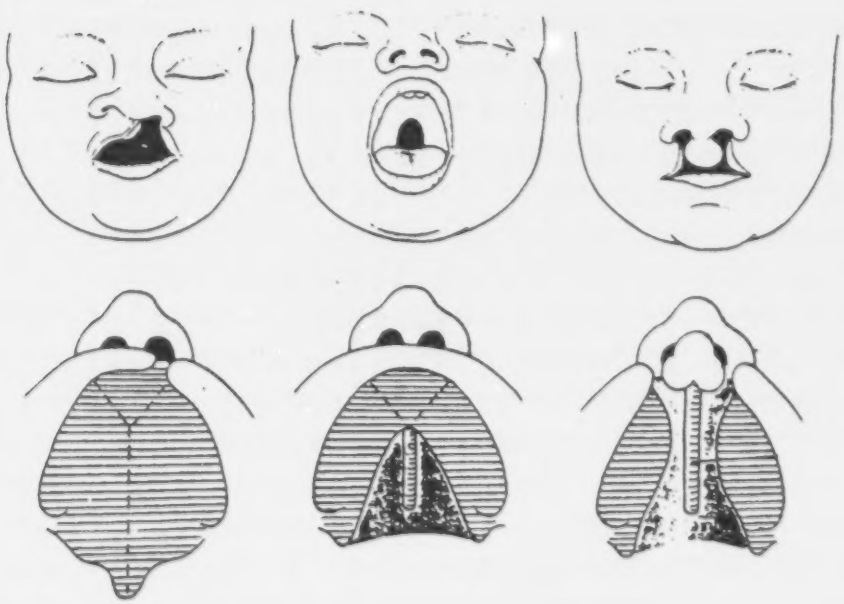
CLEFT LIP (with or without cleft palate, CL/P):
Cleft lip can be **unilateral** (UL, one-sided) or **bilateral** (BL, two-sided). Cleft lip typically occurs between the side and center (cupids bow) portions of the upper lip. It may involve only the lip, or extend into the gum and even into the primary (hard) palate.

CLEFT PALATE (CP):
Cleft palate results from a failure of fusion of the palatal shelves. Isolated cleft palate (without cleft lip) is in the back of the palate. It can involve the hard and soft palate or just the soft palate. Children with cleft palate typically have underdevelopment of the mid-face (flat midface) and often a small chin.

Submucous cleft palate: Submucous cleft palate involves the muscles (but not the bones) of the soft palate. Swallowing and speech are often affected.

Bifid uvula:
Bifid (split or double) uvula (little thingee that hangs down in the back of the throat) is considered a mild form of cleft palate. This alone does not usually create problems. It may be a sign to look more closely for a possible submucous cleft palate.



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EFFECT OF CLEFTING IN THE CHILD:

Although cleft lip is a cosmetic problem, the more immediate concern is the effect on feeding. Special nipples may help the child suck from a bottle. Nursing is sometimes difficult or even impossible. Cleft palate makes feeding even more difficult, as the milk can pass through the palate into the nose. Children with CHARGE often have additional feeding problems due to facial palsy, reflux, and/or swallowing problems (see FEEDING section). **Nurses and other specialists in a cleft palate clinic often have extensive experience with feeding difficulties and can be a big help to parents.**

Swallowing and speech are affected by cleft palate, sometimes even after surgical correction. Children with a cleft palate are more prone to ear infections and aspiration. Children with CHARGE (with or without a cleft) typically have multiple ear infections and require multiple PE tubes (see EARS section). Children with cleft palate are prone to conductive hearing loss (see HEARING section

Most children with CHARGE and a cleft (and many without clefts) are unable to take food by mouth for some time and require a gastrostomy tube (g-tube) for some time (see FEEDING section).

Children with cleft lip/palate often have orthodontic problems which will need to be addressed. Children with CHARGE may have particular dental problems as well, but these have not yet been well-described.

MEDICAL MANAGEMENT

TEAM APPROACH:

It is the recommendation of the American Society of Maxillofacial Surgery as well as the American Society of Plastic and Reconstructive Surgeons that management of the patient with facial clefting be provided by an interdisciplinary team of specialists offering a coordinated and consistent philosophy and a continuum of care. Members of the Craniofacial team may include a plastic surgeon, oral surgeon, orthodontist, otolaryngologist, ophthalmologist, social worker, nurse, audiologist, speech pathologist, and geneticist. Every institution will have a slightly different team and perhaps a different approach to facial clefts. Make use of these team members! Many of them will have had experience with feeding problems in other children.

TREATMENT/SURGERY:

The treatment for facial clefting involves a series of surgical procedures which may take place over many years, even through the patient's young adult life. The actual timing and type of treatment takes into consideration the child's growth, development, and other medical problems. Prior to surgery, it is often necessary to take dental impressions. Some children will need a pre-surgical orthodontic appliance and home therapy to prepare for surgery.

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Surgery is not usually scheduled immediately for cleft lip and palate. Children with CHARGE often have more immediate surgical needs, such as choanal atresia or a heart defect. Many palatal clefts will be repaired between eight and twelve months of age in a single stage, although surgery on very wide clefts may be delayed until up to eighteen months of age.

OTHER TESTS/PROCEDURES:

Audiological (hearing) testing should be done early. Children with CHARGE have an even higher risk of hearing loss than do other children with clefts. Insertion of PE tubes and the taking of dental impressions may be done during the same anesthesia as cleft surgery.

Non medical management
Most of the non-medical concerns related to facial clefts are feeding concerns. See the FEEDING section for more information.

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CHARGE SYNDROME: ESOPHAGEAL ATRESIA AND TRACHEOESOPHAGEAL FISTULA PHYSICIAN INFORMATION

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TYPES AND FREQUENCY OF TRACHEO-ESOPHAGEAL ANOMALIES IN CHARGE:

Tracheo-esophageal fistula	20%*
* or higher, often H-TEF is not diagnosed in the newborn period	
Esophageal atresia	15%

Related findings frequent in CHARGE which may affect treatment and/or management of tracheo-esophageal anomalies:

Polyhydramnios (prenatal)	frequent
Tracheomalacia	frequent
Laryngomalacia	30%
Gastro-esophageal reflux	50%
Esophageal dyskinesia	75%
Pharyngo-esophageal dysmotility	80%
Facial palsy	50%
Cleft palate	20%
Choanal atresia	30%
Cranial nerve IX/X anomalies	frequent
Abnormal Ba swallow	very frequent
Gastrostomy in patient w/o EA	36%
Fundoplication	frequent

DIAGNOSTIC TESTS:

- NG tube
- X-ray
- Barium swallow

The newborn with CHARGE who has TEF/EA will have copious oral secretions and a nasogastric tube will not pass into the stomach.

MEDICAL CONSEQUENCES OF FEATURES

Children with CHARGE and TEF/EA have higher mortality than other children with CHARGE. This is especially true when combined with choanal atresia and/or a heart defect.

Many individuals with CHARGE have abnormal esophagus motility and/or gastro-esophageal reflux. A significant percentage also have difficulties with suck and swallow resulting from cranial nerve anomalies. These problems further complicate feeding problems in these children. Many children need gastrostomy feeds for years.

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MEDICAL MANAGEMENT AND CAVEATS

Treatment of EA/TEF in CHARGE may be similar to treatment of isolated EA/TEF. However, there are a number of special considerations in these cases:

1. Heart defects (present in 2/3 of children with CHARGE) may be complex. These may affect timing of surgery and overall stability of the patient.
2. Possible unexpected reaction to anesthesia. Some children with CHARGE are resistant to sedation, while others are slow to recover from anesthesia. Because of risks of anesthesia, it may be appropriate to combine surgical procedures in these children.
3. Laryngomalacia or tracheomalacia is common in CHARGE and can result in surgical or anesthetic complications. The mortality rate in children with this combination of features is high.
4. Cranial nerve palsies complicate post-operative feeding in a major way. Most importantly, apparent abnormalities of cranial nerves IX and X cause secretions and food to pool in the pharynx. Esophageal peristalsis is uncoordinated. Aspiration and reflux are common. This improves in weeks, months or years. A small number of children have permanent problems. Facial palsies are usually unilateral and may affect lip closure.

NON-MEDICAL MANAGEMENT

A feeding specialist is a must for children with CHARGE with TEF/EA. All of them will have feeding difficulties, which are likely to last for years. Transition from gastrostomy feed to oral feeding can take years, as can transition from pureed to solid and liquid foods. Growth may be impaired by poor caloric intake.

If a patient has CHARGE, TEF/EA is likely to be only one of a number of serious medical problems for that child. It is critical to talk to the parents about the whole child. An orofacial team which includes ENT, audiology, and feeding specialists can be a great help to parents because they have experience with feeding difficulties, PE tubes, and ear infections, all of which are common in CHARGE. Communication with other specialists following the child is critical to effective care of these children. These children often have multiple medical appointments every month or even every week, so coordination of medical appointments is appreciated by parents.

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CHARGE SYNDROME: ESOPHAGEAL ATRESIA (EA) AND TRACHEOESOPHAGEAL FISTULA (TEF): PARENT INFORMATION

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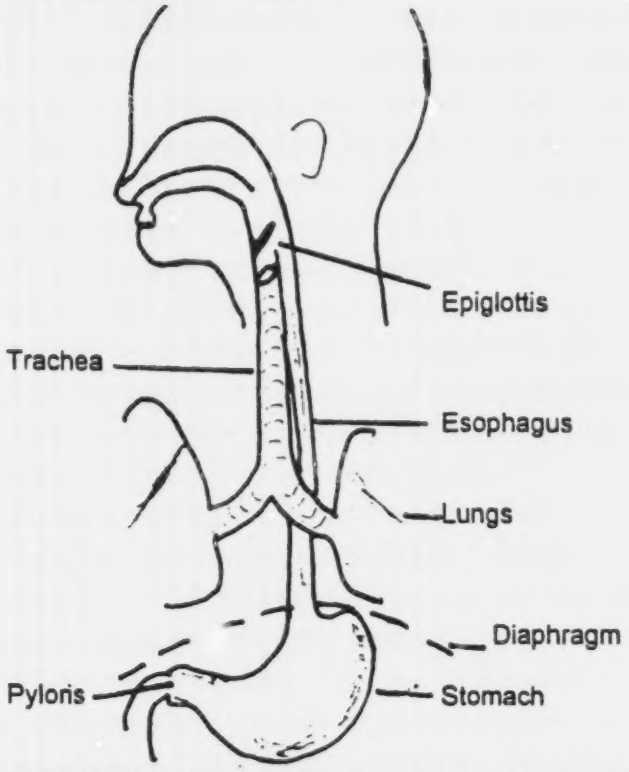
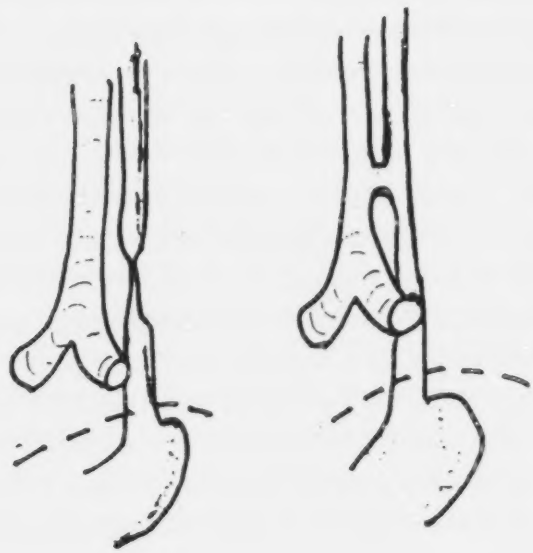
Structure and function





Esophageal Atresia

TEF (H)

The **epiglottis** is a small flap in the back of the throat. The purpose of the epiglottis is to ensure that food goes down the esophagus and air goes down the trachea.

The **trachea** (windpipe) is the tube leading from the back of the throat to the lungs for breathing. The trachea runs along side the esophagus and then splits in two along either side of the esophagus, leading to the two lungs. The trachea is surrounded by rings of cartilage and muscle.

The **esophagus** is the tube leading from the back of the throat to the stomach. It is surrounded by a coat of muscle, which help the food move down into the stomach. Food normally moves down the esophagus into the stomach. Normally, when food or saliva is swallowed, waves of movement in the esophagus (peristalsis) propel the food into the stomach. The opening from the stomach to the intestines is called the **pyloris**. Swallowing normally begins in the first three months of prenatal development.

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Problems associated with CHARGE

Esophageal atresia (EA) refers to an esophagus which ends blindly before reaching the stomach. It is often found along with tracheo-esophageal fistula. Often, polyhydramnios (excessive amniotic fluid) was noted during pregnancy. Prenatally, the fetus normally swallows and recycles amniotic fluid. If the fetus cannot swallow due to EA, polyhydramnios can result. Also, the stomach may not be visible by ultrasound exam because it is not filled with amniotic fluid.

Once the baby is born, he or she will often have copious frothy secretions pouring out of their mouths which requires constant suctioning until the EA is treated. A baby with EA cannot eat by mouth (food can't get to the stomach) until the atresia has been surgically corrected.

Tracheo-esophageal fistula (TEF)
A fistula is a connection. TEF is a connection between the esophagus and the trachea. This connection allows food (from the esophagus) to enter the lungs (aspiration). Food in the lungs can cause pneumonia (referred to as aspiration pneumonia), which can be very serious. If there is a TEF with EA, all the food taken in will end up in the lungs, as the esophagus does not connect to the stomach, but ends blindly.

The hardest type of TEF to diagnose is **H-type TEF**, where the esophagus does not end blindly, but there is a fistula or connection between the trachea and the esophagus. In H-type TEF, air can enter the stomach (from the trachea) and food can enter the lungs. Air in the stomach can cause bloating of the baby's abdomen and can make the baby very uncomfortable

Symptoms of TEF/EA
If you had polyhydramnios or the fetal stomach could not be seen on ultrasound, TEF/EA may have been suspected during your pregnancy. After a baby is born, esophageal atresia may be suspected in the first few days if the baby doesn't tolerate feeding (chokes and spits), doesn't seem to swallow saliva (froths at the mouth), seems to aspirate the milk into his or her lungs, or has abdominal distention (bloated tummy).

H-type TEF may not be recognized until much later. The typical symptoms are choking with feeding, excessive gas in the stomach or intestines, and **frequent aspiration pneumonias.**

By now, you probably already know enough about CHARGE to recognize that many of these symptoms can also be caused by other problems associated with CHARGE. Choanal atresia, cleft palate, reflux, and cranial nerve abnormalities can all interfere with breathing and eating. Infants with CHARGE may not be fed right away due to other problems. And so on. This means that TEF and associated problems may not be diagnosed as quickly or as thoroughly in infants with CHARGE.

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Diagnosis of TEF/EA
Esophageal atresia is usually diagnosed by placing a tube either down the nose or through the mouth, down the throat and into the stomach. An X-ray is taken to document that the tube is in the esophagus (not the trachea) and that it has reached the stomach. If the baby has EA, instead of ending up in the stomach, the tube will curl up in the blind-ending pouch. If there is a tracheo-esophageal fistula (which is very common with EA), the tube may end up in the lungs.

Other tests which might be done to diagnose or confirm the diagnosis are bronchoscopy or esophagoscopy. Barium swallow may be needed to diagnose an H-type TEF, where the esophagus is attached to the stomach, but there is a connection to the trachea.

Treatment
EA and TEF can be surgically repaired. Before the repair and while it is healing, the baby will need to be fed by a gastrostomy tube or button. This is a tube or opening which goes directly into the baby's stomach, bypassing the esophagus. How long the baby will require the G-tube is variable. Many children with CHARGE are fed by g-tube for years (see below and Feeding section)

Occasionally, EA will be repaired and yet a small H-type TEF is not recognized. The remaining TEF can still cause multiple aspiration pneumonias. A barium swallow test might be helpful in diagnosing the TEF.

Outcome and complications:
Even after surgical correction, there can be some leakage at the site of the reattachment. The esophagus can tighten up. If this happens, it can be treated by dilating the esophagus. Many children with CHARGE have swallowing difficulties, gastroesophageal reflux, recurrent pneumonias, and poor growth. Some of this may be due to TEF/EA, but much of it may be due to other complications of CHARGE (see FEEDING section) Once the baby has recovered from surgery, oral feeding might be tried. However, individuals with CHARGE typically have other features which complicate oral feeding. Be careful not to push it!

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THE AIRWAY IN CHARGE – PHYSICIAN SECTION

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Airway management is often one of the major problems in CHARGE. Many of the anomalies are well-known entities with well-established management protocols. However, CHARGE has several fairly unique features which make decision-making difficult, particularly whether or not to do a tracheotomy.[1,2]

ANOMALIES

Choanal atresia – surgical management is not different from other conditions.[3]

Laryngo-tracheo-bronchomalacia – can be severe[4] leading to collapse of bronchi and decreased air movement.

Cleft lip and palate – surgical management is standard but feeding management is not. See gastroesophageal reflux below.

Gastroesophageal reflux related to neonatal brain stem dysfunction, which is the major cause of feeding problems in CHARGE. NBSC, previously reported in children with Pierre Robin sequence is a group of four types of symptoms involving the supranuclear region of the IXth, Xth and XIIth cranial nerves.[5,6,7] These symptoms, important in the first two years of life, seem to be related to a developmental defect of the suck-swallow central pattern generator in the solitary tract.[8] Recurrent aspiration pneumonia due to this problem is a major cause of morbidity and mortality.[9]

Heart disease – anomalous pulmonary venous return and vascular rings have been reported.[10]

Tracheoesophageal fistula – surgical management is the same but, again, feeding is not.

Other more minor airway anomalies and those above which are not recognized prior to anesthesia can lead to difficulty with intubation.[11]

Recent anecdotal report of asymmetry of diaphragm movement was noted on fluoroscopy of a one-year old.

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MANAGEMENT DECISIONS

Tracheotomy: While most physicians try to avoid tracheotomy in these patients, the swallowing problem can be so severe that the child does not swallow its own secretions. The secretions, then, become copious and are a management problem in themselves. They obstruct the upper airway and may be aspirated along with any feedings. After tracheotomy is performed, secretions and any formula or food is usually seen coming out the trach with suctioning. Tracheotomy may be performed in a child who has NBSC until such time as it resolves spontaneously usually over several years. Rarely is there a need for permanent tracheotomy, but several cases are known.

Tracheal diversion: This procedure has been performed in older children whose NBSC has not resolved spontaneously, are unlikely to develop speech, and who show a strong desire to eat.

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SWALLOWING AND FEEDING IN CHARGE SYNDROME
For the Physician

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ANOMALIES SPECIFIC TO CHARGE

Swallowing difficulties, one of the major features of CHARGE syndrome, particularly in the first years of life, occurred in 96% of ourchildren with CHARGE[1]. Other authors report between 31and 88%.[2,3] The main causes are as follows:

Poor suck-swallow coordination. Neonatal Brain Stem Dysfunction (NBSD):

Neonatal brain stem dysfunction is the major cause of feeding problems in CHARGE. NBSC, previously reported in children with Pierre Robin sequence (4-6), is a group of four types of symptoms involving the supranuclear region of the IXth, Xth and XIIth cranial nerves. These symptoms, important in the first two years of life, seem to be related to a developmental defect of the suck-swallow central pattern generator in the solitary tract.[7]

(i) sucking and swallowing skills disorder,
(ii) esophageal dyskinesia,

Clinically, both of these symptoms result in poor suck, prolonged feeding time, milk aspiration, unexplained cries during bottle feeding, nasal reflux, regurgitation and ALTE (Apparent Life Threatening Event) during feeding. They may lead to pharyngeal congestion, aspiration pneumonia and failure to thrive. The esophageal dyskinesia is responsible for gastroesophageal reflux (GER) that is poorly managed by classical medical treatments.

(iii) glosso-pharyngeo-laryngomalacia
(iv) sympathetic-parasympathetic cardiac rhythm dysregulation.

The glosso-pharyngeo-laryngeomalacia is responsible for obstructive apneas or hypopneas and subconsequent hematosis while the vagal dysregulation may result in ALTE, induced by all previously noted symptoms. Sucking and swallowing incoordination of NBSD resolves when corticalization of feeding occur, after 6 mo. of life.

Facial palsy

The facial nerve plays a role in face and lip movements which increase sucking problems. Facial palsy in CHARGE is almost always unilateral and peripheral due to dysgenesis of the VIth nerve extra bulbar pathway. Determining the origin of facial palsy is important for prognosis and electromyogram may help. Facial palsy may show little or no improvement with age and may be responsible for further aesthetic problems.

Malformations of esophagus, larynx or choanae ; cleft lip and palate.

Anatomical malformations of the organs involved in sucking, swallowing and breathing are also responsible for swallowing and are discussed in other sections of this manual.

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Hyposmia

Abnormal olfactory lobes have been noted in CNS autopsy of CHARGE patients[2] and recently, the common embryological origin of olfactory tracts and hypothalamus involved in hormonal defects (LHRH) led to a better MRI analysis of that brain region, showing frequent anomalies.[8] personal data

Even at birth smell seems to be involved in feeding behavior and increases appetite. This is more evident after corticalization of feeding, i.e. at the end of the first year. The clinical consequences of hyposmia need to be further defined, even in young infants, by perfecting olfactometric scales for children before language acquisition. Olfactory competence in CHARGE has been poorly investigated for both technical and medical reasons: i) olfactometric scales requiring good language cannot be performed by a normal child before 10 years of age and ii) hyposmia is a minor problem that escapes notice.

Exogenous factors

Finally, feeding difficulties may also be secondary to exogenous factors such as cardiac or pulmonary dyspnea, effects of initial nasogastric tube feeding whatever the reasons of the enteral nutrition and deleterious effects of the long initial hospitalization both on the child and on altering the precocious mother-child emotional bonds.

DIAGNOSTIC TESTS

Sucking and swallowing disorders are mainly evaluated by clinical means. A guided anamnesis, a good history and observation of the child during feeding are the best tests. Cineradiography may be dangerous and is not sensitive enough in mild cases. It may help determine when to restart an oral feeding program when aspiration risks diminish. Succimetry may be interesting but, in our hands, remains a research procedure.

Neonatal brain stem dysfunction can be investigated by its peripheral effects.

i) Esophageal dyskinesia may be investigated by an esophageal manometry, which provides specific information on abnormal esophageal motor control: hypertonia or achalasia of the lower esophagus sphincter, abnormal coordination of pharyngeal contraction and the upper esophageal sphincter. These features are not constant (95% of investigated children in our series had at least one abnormal manometric criterion, 60% a specific anomaly and the rest a less specific abnormal motility of the esophagus). Investigations of the neurologically-based GER are only necessary when surgical treatment is considered. X-ray barium transit is useful but pH meter readings are less so since results may be normal even if vomiting is evident.

ii) Laryngeal endoscopy may show specific aspects of hypotonia of the tongue base, pharynx walls and larynx. It may also show salivary stasis and peptic inflammation of the larynx and trachea secondary to gastric acid reflux and feeding aspirations.

iii) A 24-hour electrocardiogram recording (Holter) with ocular compression test may help to evaluate a vagal hyperactivity.

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iv) Evoked potentials of the brain stem may show abnormal delay in tracing, particularly during the first steps of the auditory evoked potentials (AEP) used in assessing hearing loss. Swallowing disorders by themselves do not require this investigation in practical terms.

v) Facial and endobuccal electromyography (EMG) as well as dynamic EMG (when it is not dangerous) during bottle feeding may be important to determine the origin of cranial nerves defects. Most often, recording of the 7th cranial nerve shows peripheral palsy. Recordings of 9th 10th and 12th nerves separately are normal but dynamic EMG during bottle feeding shows poor coordination between muscles innervated by the 9th and 10th nerves respectively.[9] This test mainly has theoretical interest but it is quite aggressive and needs a specialized operator.

vi) Finally, investigations of olfactory abilities may be performed with adapted scales (personal data) or by MRI of the olfactory lobes. Again, these investigations are not required for proper treatment of swallowing difficulties.

MEDICAL CONSEQUENCES

Medical consequences of swallowing disorders are numerous. They alter pulmonary status, induce chronic bronchitis with a risk of hypoxemia and hypercapnia and adversely affect cardio-pulmonary vascularization, quality of sleeping and psychomotor development. Feeding aspirations and stasis in the pharynx worsen respiratory obstructive syndrome. Discomfort and pain induced by swallowing disorders lead to a decreased intake and failure to thrive. Sucking and swallowing disorders alter mother-child relations and increase the risk of further anorexia even when organic problems are solved.

Personal data. 30 children with CHARGE from 1 to 9 years

- Adapted diet and feeding procedures only — 3 children
- Nasogastric tube only — 7 children (3 precocious deaths)
- Gastrostomy and GER surgical treatment — 20 children
- Mean age of artificial nutrition weaning — 3 years (min 16 mo, max 7 yr)
- Mean age of normal eating recovery (if reached): — 5 years
- Number who do not eat after 6 years of age — 2 children

MEDICAL AND NON MEDICAL MANAGEMENT *primum non nocere*

Apart from surgical treatment of esophagus and upper airway anatomic malformations, swallowing disorders have no radical medical therapy. Spontaneous resolution of functional disorders of sucking and swallowing is generally good and the major challenge is to wait for their natural improvement in the best conditions. One should recognize feeding disorders early in order to prevent deleterious consequences, reduce all contributing factors, teach feeding by multisensory and physiotherapeutic means and support parents' psychologically:

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- Ask the mother for sucking skills disorders, even in the neonatal period:
- Improve sucking and swallowing mechanic, when it is not too poor, by using soft and largely perforated nipples, thickened milk, small meals and avoiding forced feeding.
- If swallowing disorders have respiratory or growth consequences, enteral nutrition is required, either exclusively if aspirations are present or as a night supplement when possible. Enteral nutrition avoids the respiratory consequences of feeding aspiration and induces proper weight gain.
- The choice between nasogastric (NG) tube and gastrostomy depends on the age of the child and the severity of the symptoms. Initially, NG tube feeding is started but, after few months or earlier if the procedure is not well tolerated. If the troubles persist, NG tube should be changed to a gastrostomy combined with anti-reflux surgery.
- Good airway clearance is essential to allow swallowing. Trachoeostomy is often necessary and does not alter swallowing. On the contrary, it provides better ventilation and allows tracheobronchial drainage.
- Non medical management of feeding teaching is detailed in the parents' section.

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SWALLOWING AND FEEDING IN CHARGE SYNDROME – PARENTS

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NORMAL STRUCTURE AND FUNCTION

Feeding is both one of the more essential and the more complex of the mammalian functions. Feeding of children can be divided into two periods :

1. The sucking and swallowing reflex period lasts from fetal life to the age of 6 months and essentially requires the good organization of sensory and motor pathways going and coming from the face and the mouth to the brain stem.

2. The corticalized feeding period, lasting from the middle of the first year to adulthood, involves voluntary processes for the feeding orally and are influenced by sensory, psychological and environmental factors. Adult feeding processes are matured between 2 and 3 years of age.

Moreover, appetite and caloric intake are regulated by several neural mediators coming from the intestinal tract and the CNS (hypothalamus, thalamus, cortex). For children with CHARGE syndrome, the main problems seem to occur during the first period, during the brain stem development and organization. We do not know specific treatment for these disorders so the aim is to wait for the second period when feeding improves on its own.

TESTS USED AND SPECIALISTS

Swallowing disorders are generally very well evaluated by the mother. From their child's first days of life, mothers may observe poor sucking, inability for the baby to be breast fed, discomfort with bottle, unexplained cries, bottle refusal or prolonged feeding time, skin color changes during feeding, breathing difficulties or cough increased by feeding, frequent spitting up, nasal reflux or poor weight gain.

The only two obligatory investigations are, in my opinion, a chest x-ray to detect pneumonia aspirations and an examination of the airway by a competent pediatric otorhinolaryngologist (ENT).

Other investigations may be useful to characterize the origin, the mechanism and the severity of the symptoms. They are performed differently depending on the child and the medical team (see doctor section).

MANAGEMENT

Swallowing difficulties are as badly tolerated by the child as by his/her mother. Young infants affected with sucking, swallowing and breathing difficulties are not able to grow, to develop and to explore their environment in a proper way. Moreover, swallowing and feeding disorders cause great parental anxiety, for both medical and psychological reasons.

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Parents have an important role regarding swallowing difficulties in pointing out the signs to the doctor. Parents need to understand that what appear to be very drastic and aggressive techniques (nasogastric (NG) tube, gastrostomy, tracheostomy and no oral feedings) would improve their child's comfort. These medical options are not easy to accept, especially when the problems do not seem to be major ones. Performing surgery may make their child seem more gravely affected and give a feeling of failure to the caretaker. The advantages of these interventions are observed after they done. Discussing options with other parents of children with CHARGE or looking at other children who have had these procedures may help the families during these periods.

Parents also have a great role in the non-medical management of these difficulties by learning how to prevent worsening of the feeding problems and learning early how to adapt the teaching of a normal feeding by working with a Feeding Team. First by their positive attitude without guilt that they are at fault, no forced feeding and no excessive anxiety. Second by their presence and involvement in medical care, using adapted bottles, breast exclusion, by handling, massage or any methods to keep close physical contacts with the baby.

Learning to feed by mouth
After several months of poor coordination of sucking and swallowing, children may be frightened by food. The first step consists of avoiding bad experiences with food. Non-nutritive sucking with a pacifier is useful. Then, the child must get acquainted again with his feeding instrument, that is to say his face, lips, gums. These vulnerable areas have to be approached with soft massage, by mouth games, etc. and always with close contact with the child. The pleasure of feeding has to be taught using pleasant and intense smells (even if the child's sense of smell seems to be poor), colors or tastes.

Food with a smooth texture should be given with a soft plastic spoon. Problems tolerating solid food can last several years. From the end of the first year, solid pieces may be offered to the child, allowing himself the peace from his hand to his mouth, in a totally voluntary procedure. After the period of aspiration risk, drinking must be taught too, playing with pure water in a glass. Chewing must be stimulated too by elastic texture, placed in the lateral part of the mouth.

The management of feeding problems is a subjective procedure, depending on team and culture and requiring multidisciplinary interveners. In our opinion, this management must be a medical concern from the first week of life of a children affected with CHARGE syndrome.

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CARDIOLOGY IN CHARGE SYNDROME

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TYPE AND FREQUENCY OF CONGENITAL HEART DEFECTS (CHDS) IN CHARGE

- 75% - 85% have a CHD
- Almost every type of CHD has been described, including "typical" VSD, ASD, PDA. Many children have multiple, complex congenital heart defects.
- There are proportionally more conotruncal and aortic arch CHDs (30-40% of those with a CHD)

Type of CHD (below)	Lin	Wyse	Tellier	Round #
# patients studied *	53	50	40	
Conotruncal, aortic arch (1)	42%	47%	33%	30-40%
Simple shunts, R/L obstruction (2)	32%	50%	57%	40-50%
AV/endocardial cushion (3)	15%	10%	10%	10%
Miscellaneous (4)	10%		5%	5%

* many had more than one CHD

(1) Conotruncal: tetralogy of Fallot, double outlet right ventricle, aberrant subclavian artery, right aortic arch, truncus arteriosus, interrupted aortic arch (type B), conoventricular VSD.

(2) Shunts: atrial septal defect, ventricular septal defect, patent ductus arteriosus, pulmonic stenosis/atresia, tricuspid stenosis/atresia, aortic stenosis, mitral stenosis, coarctation, hypoplastic left heart syndrome.

(3) AV canal: complete atrioventricular canal, atrial septal defect, primum.

(4) Misc: complex single ventricle, anomalous pulmonary venous return, others not specified.

DIAGNOSTIC TESTS AND CAVEATS

1. Echocardiography . In addition to intracardiac anatomy, imaging should look for aortic arch anomalies, such as vascular ring and aberrant subclavian artery.
2. Catheterization provides additional information about pressures and anatomy. In one study, 1/4 of the children with CHD also had renal anomalies. Although renal ultrasound and IVP/VCUG are more definitive, delayed abdominal films at the time of a cath may be useful in screening for urinary tract malformations.
3. Electrocardiogram (ECG) to study electrical activity. May be supplemented by longer term Holter monitoring.
4. Occasionally: treadmill stress testing, MRI scanning.

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MEDICAL MANAGEMENT WITH CAVEATS

1. In one study of 50 CHARGE patients with CHD, 75% required surgery.
2. Children with CHARGE may be resistant to chloral hydrate sedation.
3. Anesthetic risk is increased in children with airway involvement such as **choanal atresia**, or **laryngotracheomalacia** (both common in CHARGE). Children with choanal atresia and complex heart defects have the highest rate of serious complications and/or poor outcome.
4. Swallowing problems with increased secretions (presumably due to involvement of cranial nerves IX and X) may present an additional risk of aspiration.
5. Prostaglandin administration may be risky because of a high mortality in CHARGE following prostaglandin administration.
6. Hypocalcemia may be due to the absence of the parathyroids in the small number of CHARGE children with classic DiGeorge sequence. However, hypocalcemia may also by due to hypoparathyroidism in other patients.
7. Heart problems are only one component of growth failure. Others include feeding problems, frequent illnesses (especially chronic otitis media and respiratory infections), and possibly growth hormone deficiency.

NON-MEDICAL ISSUES

Many children with CHARGE syndrome have dual sensory impairment (hearing and vision loss) in addition to the medical problems. This combination (medical concerns along with sensory impairment) has a tremendous impact on development. All of these children will be developmentally delayed. With appropriate medical and educational intervention (including a deafblind specialist), many of these children will eventually function in the normal to above normal range of intelligence. Do not assume mental retardation based on early developmental delay.

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CARDIOLOGY: THE HEART IN CHARGE for Parents

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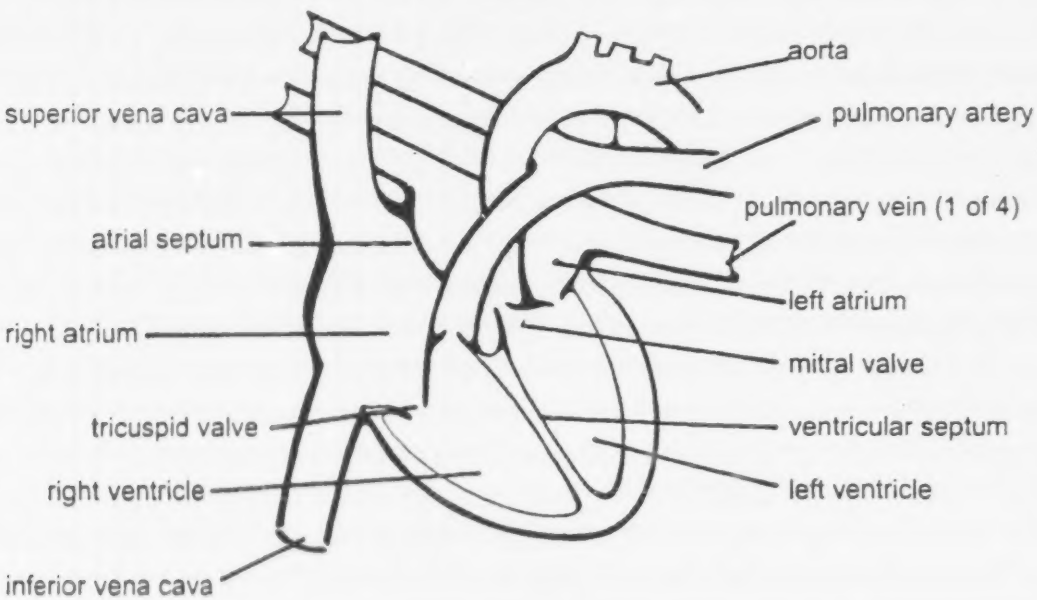
NORMAL STRUCTURE AND FUNCTION OF THE HEART

The heart is a muscular pump (myocardium) with inflow & outflow pipes (blood vessels). It can be thought of as a house with:

- Two upper rooms (right atrium [RA], left atrium [LA])
- Upstairs dividing wall (atrial septum [AS])
- Two lower rooms (right ventricle [RV], left ventricle [LV])
- Downstairs dividing wall (ventricular septum [VS])
- Two one-way swinging doors between upstairs and downstairs rooms (tricuspid valve, mitral valve)
- Large pipes entering upper right room from body (superior vena cava, inferior vena cava)
- Smaller pipes entering upper left room from lungs (pulmonary veins)
- Two large pipes, one exiting each ventricle (pulmonary artery, aorta)
- Electrical system (conduction system)
- Internal plumbing (coronary arteries) which supply blood to the heart muscle itself



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BLOODFLOW IN THE NORMAL HEART:

1) Blue blood returns from the body through veins to the **superior and inferior vena cava**, which empty into the **right atrium**. This blood passes through the **tricuspid valve** to the **right ventricle**.

2) The right ventricle pumps the blood through the **pulmonary valve** into the **pulmonary arteries**, which carry the blue blood to the lungs. The blood picks up oxygen in the lungs and turns red.

3) Red blood returns from the lungs through the **pulmonary veins** to the **left atrium** of the heart. From the left atrium, the blood passes through the **mitral valve** into the **left ventricle**.

4) From the left ventricle, the blood is pumped through the **aortic valve** into the **aorta** and from there throughout the body.

TYPES OF CONGENITAL HEART DEFECTS (CHD)

A. Holes in dividing walls, which can allow blue and red blood to mix

ASD: atrial septal defect (hole between upper rooms)

VSD: ventricular septal defect (hole between lower rooms)

AV canal/cushion defect: atrioventricular septal defect (large hole between upper rooms and lower rooms, including a hole in the floor)

B. Major plumbing problems with pipes, which can transport blood to incorrect chambers or restrict flow of blood through a vessel : ** These are very common in CHARGE

Truncus: arteriosus (1 pipe instead of 2 leaving the lower rooms)

Transposition: transposition of the great vessels (pipe positions switched leaving the lower rooms)

ToF: tetralogy of Fallot (1 pipe narrow, 1 pipe overriding, with hole in wall)

DORV: double outlet right ventricle (both pipes exiting leaving the right lower room)

Coarct: coarctation of the aorta (narrow pipe heading towards the body)

Interrupted aortic arch (pipe flow cut off heading towards the body)

TAPVR: total anomalous pulmonary venous return (pipe hooked to wrong room)

C. Leaky valves, which can allow backflow of blood through the doorway

Tricuspid or mitral regurgitation (backflow between lower and upper rooms)

Pulmonic or aortic regurgitation (backflow into lower room from pipe)

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D. Tight valves, which can restrict flow

Tricuspid or mitral stenosis (narrowing of doorways between upper and lower rooms)

Pulmonic or aortic stenosis (narrowing of pipes)

E. Abnormal rooms

Single ventricle (one large lower room)

HLH: hypoplastic left heart syndrome (very small lower left room)

Tricuspid atresia (small lower right ventricle)

CHDs in CHARGE syndrome

How common are heart defects in CHARGE?

Approximately two-thirds (60-80%) of children with CHARGE have a CHD. Many, but not all, of them are serious.

Is there a typical pattern of heart defects in CHARGE?

Any heart defect is consistent with CHARGE, but ToF, DORV, and VSD are especially common. Some children with CHARGE have extremely complex heart defects that do not easily fit into just one category.

Are the typical heart defects mentioned unique to CHARGE? No, they are also common in:

VCFS/DiGeorge complex/del22q11
Hemifacial microsomia/Goldenhar syndrome
Retinoic embryopathy

DIAGNOSTIC TESTS WHICH MIGHT BE DONE

Chest x-ray

Electrocardiogram (EKG), Holter monitoring

Echocardiogram (echo, ultrasound)

fetal/prenatal

postnatal

Cardiac catheterization (cath)

Exercise test (stress test)

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MANAGEMENT OF CONGENITAL HEART DEFECTS

Medication

- digoxin, to help the heart pump stronger
- diuretics, to get rid of extra fluid
- antibiotics, to prevent infection
- anticoagulants, to thin blood

Surgery

- to repair major plumbing problems (truncus, interruption of the aorta, ToF, DORV)
- to close holes in walls (ASD, VSD, AV canal)
- to repair loose valves (regurgitation)
- to repair tight valves (stenosis)
- to increase aorta blood flow (coarctation)

OUTCOME OF CHDS IN CHARGE

Medical outcome

Some heart defects can be totally repaired by surgery, while others can only be improved. Some children will end up with no heart problem at all, some will be much better, and others will continue to have problems with their heart. A few children with CHARGE will remain medically fragile for a long time, in part due to the heart defects.

The outcome and risks associated with heart surgery in CHARGE depend on the type of defect, the type of surgery, and on the presence of other serious health problems, especially choanal atresia tracheo-esophageal atresia, or cleft lip/palate. Many children with CHARGE have unusual reactions to anesthesia.

Developmental effects of heart defects

Remember that any child who is in the hospital for an extended period of time or who undergoes many procedures is under great stress. One outcome of the stress is delayed development. Some of the early delayed development in children with CHARGE may be attributed to multiple hospital stays and multiple surgical and diagnostic procedures. Some children actually lose milestones while hospitalized, only to regain them later. Do not be discouraged by early developmental delays, however extreme. Many children with CHARGE are truly "delayed" and will catch up over the years to come.

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MUSCLES AND BONES IN CHARGE SYNDROME
PARENT INFORMATION

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NORMAL STRUCTURE AND FUNCTION

The musculoskeletal (MS) system consists of over 200 bones and 500 muscles. A detailed description is beyond the scope of this manual. (Besides, I can never remember them all.)

Two aspects of the MS system are critical: **structure** and **function**. Normal **structure** means that all of the components of the system (that is the muscles and bones) are present and in the proper relationships to one another. Normal **function** means that the bones are maintaining the normal structural integrity of the skeleton and the muscles (through contraction) are able to move the portions of the skeleton that are supposed to move (the joints). All **structural** abnormalities lead to **functional** abnormalities (although the severity of the abnormality may not be clinically significant), but not all **functional** abnormalities lead to **structural** abnormalities. An example of this would be a person who suffers a spinal cord injury. The muscles and bones are completely normal, but the muscles are unable to function because of the absence of nerve signals.

Two other important concepts are **strength** and **tone**. Most people understand strength, but tone is a harder concept to grasp. **Strength** is the ability of a muscle, or group of muscles to work against a load. It can be objectively measured. **Tone**, however is a subjective assessment of muscle function at rest. When we are resting (that is not moving a particular muscle or group of muscles) our muscles are not completely at rest. There is a baseline level of activity that can be detected, but is not easily measured. We refer to this as **muscle tone**. Muscle tone helps with posture and maintaining normal skeletal relationships. Tone can be normal, low or high. Low tone is also called **hypotonia**. Individuals with low muscle tone are often described as floppy. This can lead to slumped posture, problems with head control, standing, etc. High tone is called **hypertonia** or **spasticity**. Individuals with spasticity feel like the muscles are constantly contracting. This can lead to abnormal joint positions and result in toe-walking, hip and knee flexion contractures, etc. An experienced physician or developmental therapist can assess Tone, but there is no way to objectively measure muscle tone. Abnormalities in muscle tone can be due to abnormalities of the **nervous system** (brain, spinal cord, spinal nerves, peripheral nerves or connections between the nerve and the muscle), or to abnormalities within the muscle itself (**myopathy**). It may be difficult to distinguish between these two causes on clinical examination.

TYPES OF MUSCULOSKELETAL ABNORMALITIES IN CHARGE

Between 30 and 50% of patients with CHARGE are born with some type of skeletal abnormality. Severity can range from clinically nonsignificant (minor changes of the creases of the palms due to short hand bones), to quite severe (missing fingers). Several patients have been reported to have fusion of fingers or toes (syndactyly) or clefting of the hand or foot.

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Low muscle tone (**hypotonia**) is very common in children with CHARGE, especially in the upper body (trunk). There have not been any patients with CHARGE known to have hypotonia due to a myopathy (abnormality of the muscles themselves). Unless new information becomes available, it is probably safe to assume that the hypotonia is due to a central nervous system abnormality (i.e. brain). Low muscle tone may have an effect on development: if the upper body is floppy, it will be more difficult to sit alone or stand. Combine weak tone with vision loss and balance problems, and you may have a child who does not walk until age 5 or 6.

Scoliosis (curvature of the spine) is common in children with CHARGE. Although scoliosis is generally thought of as a teenage problem, it has been seen in young children with CHARGE. By the teenage years, a majority of individuals with CAHRGE may have some scoliosis. This may be due, at least in part, to the low muscle tone in the upper body.

DIAGNOSTIC TESTS

The most important diagnostic test in very young children is a careful physical examination of the musculoskeletal system. Imaging studies (such as X-ray, ultrasound) are indicated if there is suspicion of an anomaly on physical examination. Skeletal survey (to look at all the bones of the skeleton) is not routinely indicated. Tests of muscle (muscle biopsy, electromyogram (EMG)) are generally not indicated, unless a primary muscle problem is also suspected.

In older children, regular physical exam for scoliosis is indicated. If scoliosis is suspected, the doctor may order X-rays to determine the extent of the scoliosis.

MANAGEMENT AND OUTCOME OF MS ANOMALIES

Medical and/or surgical management is based on the type of anomaly. They are not managed differently whether or not the child has CHARGE. Outcome following intervention is generally good, but clearly depends on the severity of the problem and the treatment that is required. Occupational therapy (OT) and physical therapy (PT) may be helpful in dealing with low muscle tone.

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GROWTH IN CHARGE - PHYSICIAN INFORMATION

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Growth in children with CHARGE has special considerations in each of the three phases: Infant Phase, Childhood Phase, Pubertal Growth Spurt.

1. ***Infant Phase:***
   At birth, children with CHARGE Association usually have normal weights and lengths.[1,2] However, within the first 9 months of life there is a decline away from the normal growth curve, down to and often below the 3rd percentile. This pattern of growth may be related to repeat hospitalizations, poor feeding, major acute illnesses such as pneumonias and multiple surgeries. The infant phase of growth is mainly determined by nutrition and those children with CHARGE who maintain their weight in the early months are often the ones who have had major nutritional intervention, usually in the form of entral feeding.[1]

2. ***Childhood Phase:***
   Although the growth rate is slower, the childhood phase of growth is the main determinant of final height as it lasts longer than the other phases. Adequate nutrition is important in this phase but also hormones play a role (thyroxine and growth hormone). Growth hormone deficiency is rare but there is an increased incidence in CHARGE.[3,4] Pre-adolescent children may have a more normal rate of growth although poor growth in infancy and lack of catch-up growth during childhood often result in a mean height at or below the 3rd percentile.

3. ***Pubertal Growth Spurt:***
   Growth deceleration associated with a delay in puberty occurs in more than 70 percent of children with CHARGE and this needs to be monitored closely. Growth in puberty involves the interaction of the sex steroids, especially testosterone and estrogen associated with growth hormone. There is anecdotal evidence that adolescents with CHARGE have fusion of their epiphyses at a later age and therefore often continue to grow in their early 20's.

The majority of children with CHARGE association have normal results on growth hormone stimulation testing. Arbitrary treatment with growth hormone in patients who have normal growth hormone levels has not been adequately studied and is probably not advised.

Nutrition **"Growth and Nutrition are Closely Related"**

Children need an adequate quantity and balance of food for optimal growth and development. CHARGE infants who have excessive surgery and a decrease in nutrition intake may require catch-up growth and their energy requirements may be 150 to 200 kcal/kg/d.

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Reference Values for Energy and Protein Requirements		
Age	**Energy kcal/kg/d**	**Protein gm/kg/d**
0-6 months	115	2.2
6 mts.-6 yrs.	95	1.8
7 yrs-10 yrs.	75	1.2
11-16 yrs.	60	1

FEEDING ISSUES IN INFANCY AND EARLY CHILDHOOD

Symptoms and Behaviors of Swallowing Problems (Also seeSwallowing Section):

1. The feeding history may include:
   Cough and choking, nasopharyngeal reflux, nasal congestion, food suctioned from the nose or from the tracheostomy.

2. Chronic chest difficulties.
   Recurrent pneumonias, apneas, frequent upper respiratory tract infections.

3. Other behaviors.
   The infant may have stresses on feeding such as sweating, gasping, straining, prolong or shortened sucking pattern. In children, other symptoms may be exhibited i.e. build-up of secretion, spiking temperatures, food remaining in the mouth, choking, eyes tearing or lurching the head forward and facial stress.

More than 90 percent of children with CHARGE have difficulty swallowing foods of different textures. As a consequence parents often discontinue trying to feed their children solid foods.

Children with bilateral posterior choanal atresia often have the most severe feeding problems and may manifest significant failure to thrive.[4] In some instances, there may be obvious physiological reasons for these problems, i.e. cleft lip and palate, facial palsy, choanal atresia, tracheo-esophageal fistula. Occult and hidden anatomical abnormalities of the pharynx and larynx have also been described.[4] These need to be considered in children and infants with feeding problems. One of the most difficult chronic management problems that families have to face is gastro esophageal reflux. There is no quick fix to the problem and different centers offer their expert team.

When gastrostomy tubes are used, children run the risk of oral pharyngeal hypersensitization, meaning that they reject substances and objects in their mouths, especially feeding devices. It is therefore important to try and keep stimulating the oral pharynx even though the child is being fed by gastrostomy or jejunostomy tube. Even with aggressive therapy many children with CHARGE continue to have feeding problems into pre-adolescent and beyond. Parents also report excessive abdominal colic similar of the type commonly found in infancy which may present to school age children and beyond. The etiology of this colic is unknown but is probably organic in nature.

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General Management in Feeding a CHARGE Infant

- Positioning of infant or child
- Use of different textures to find the ones that work
- Aspiration precautions
- Aggressive management during colds
- All of the above can be guided with a feeding team which should include an occupational therapist, physiotherapist, speech therapist, behavioral psychologist, and dietician.

Breast Feeding - often difficult in CHARGE infants because of their many surgeries but not impossible especially if expressed breast milk is used.

Weaning:

Solid foods are usually introduced between the ages of 3 and 6 months. After 6 months of age, milk only can lead to anemia and deficiencies in vitamins and iron. Lumpy foods, which are usually introduced at about 6 months, are hard for CHARGE children to manage. Different textures and tastes, may also be a problem. Many families use very pureed foods until childhood.

Failure to Thrive:

All children with chronic illness can have failure to thrive. Congenital heart disease or respiratory disorders alone can be responsible for failure to thrive. In CHARGE children, malnutrition may result from a combination of decreased intake, malabsorption and increased requirements because of increased work of breathing. Malnutrition from many of the aspects of CHARGE association can respond to entral feeding.

Malnutrition must be recognized and accurately defined so decisions can be made about feeding. Evaluations are divided into assessment of past and present dietary intake, measurements and lab assessments as in the table above. The consequence of malnutrition is a multi-system disorder. Malnutrition worsens the outcome of illness, respiratory muscle dysfunction and may delay a child being weaned from a mechanical ventilator. Malnourished children are less active, less exploratory and more apathetic. These behavioural abnormalities are rapidly reversed with proper feeding.

Assessment of Nutritional Status
Anthropometry · weight · height · mid arm circumference · skin fold fitness **Lab Data** · low plasma albumin · low concentration of specific minerals and vitamins **Food Intake** · dietary recall · dietary diary

Immunodeficiency
- low lymphocyte site count
- impaired cell mediated immunity

Short Stature or Failure to Thrive:

Short stature and failure to thrive are usually defined as height or weight below the 2nd or 3rd percentile (approximately 2 standard deviations below the mean). Serial measurements are the most helpful as they allow detection of change in the rate of growth. Growth failure can be defined from the child's growth parameters falling across percentile lines plotted on a growth chart. In this way, growth failure may be identified even though the child's height and weight are still above the second or third percentile. As most children with CHARGE have a normal birth weight and length, then it is often obvious within the first 9 months when they fall away from the middle percentiles. CHARGE children who suffer from malnutrition and chronic illness are usually moderately short but significantly underweight. In contrast, if growth hormone deficiency is the cause for growth delay, children are usually somewhat overweight for their height.

Short stature may cause psychological problems. Adults assume that the children are younger than their true age and so treat them inappropriately.

Growth Hormone

Diagnosis of growth hormone deficiency is difficult because of the pulsator nature of growth hormone secretion. In pituitary provocation tests, a variety of stimuli are used to provoke growth hormone release. The most common is clonidine, glucogone and insulin induced hypoglycemia. The hypoglycemia induced by insulin is potentially dangerous. To avoid these complications, the test should only be performed in specialist centres. Growth hormone deficiency is treated with biosynthetic growth hormone, which is given by subcutaneous injection usually daily. There are other potential applications of growth hormone therapy, which is under study including children with different syndromes and those with chronic illness. CHARGE would fall within this realm, however, growth home is not being used at present for children who have not been diagnosed with growth hormone deficiency.

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FEEDING AND GROWTH IN CHARGE - FOR PARENTS

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Although some children with CHARGE have growth hormone deficiency, much of the slow growth in CHARGE is likely due to nutritional status and general medical problems.

Difficulty in feeding is one of the most common and prolonged problems in CHARGE. As a consequence of poor feeding, the long term poor nutrition results in growth failure and can contribute to the developmental delay. Assessment of growth is important and below are the growth parameters that should be measured routinely in your children (3-4 times per year):

1. Weight - If possible, measurements should be on the same weight scales.
2. Length or Height - In children under 2 years old, length is measured lying horizontally. Because of balance problems and delayed mobility, children who are not comfortable standing to be measured should also be measured horizontally.
3. Head circumferences should be measured in the first 2 years of life. This is the widest circumference around the head called the occipital frontal circumference and is an indication of brain growth and overall size.
4. The measurements of growth parameters should be plotted on percentile charts. It is wise to keep a copy in your binder of your own child's growth progression and ask to be shown how to plot and read the charts.
5. Clinic - It is important to attend a growth and/or endocrine clinic. If growth hormone deficiency is suspected, your doctor may order special testing.

Feeding and Swallowing In Children With CHARGE (Also seeSwallowing Section):
Feeding problems are frequent and represent a permanent concern for most parents. Feeding studies may show that the infant can suck normally but liquid pools in the back of the throat without passing smoothly into the esophagus. Swallowing can be uncoordinated and also lead to gastroesophageal reflux. Children are often uncomfortable, show signs of restlessness when they are being fed, they may gag and aspirate (the food or liquid goes down into the lungs). These children have a hard time feeding orally. Some can handle pureed foods but cannot tolerate liquids or solids. It is important to find the consistency and type of food that your child will tolerate. Positioning and behavior modification when feeding are also important. A team of therapists should be involved with your child who has feeding problems. A therapy team includes a speech-language pathologist, an occupational therapist, a psychologist and a physician with some interest in feeding problems. If there is a center where there is a feeding team, this center should be consulted.

ANATOMICAL ISSUES WHICH HAVE AN IMPACT ON SWALLOWING:

- Retrognathia - posteriorly placed mandible
- Micrognathia - underdeveloped mandible (jaw bone)
- Tracheoesophageal fistula (TEF) - presence of a fistula (a joining) between the tracheal and esophageal walls.
- Cleft lip and Palate - refers to a "split" or "separation" in the lip and/or palate
- Facial palsy - usually on one side but can be both sides. The side of the paralysis is the side where the eye has incomplete closure - more noticeable when the infant is crying.
- Choanal Atresia or stenosis

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- Lower Cranial Nerves Involved in Swallowing and Sucking (IX X XI) - affects sensory (feel) and motor function (activity) involving sucking and swallowing.

TESTS USED TO EVALUATE SWALLOWING:

Barium Swallow:
Assessment of anatomical structure function
Examines the esophagus pharynx, and larynx
Uses large amounts of liquid purees, and solids
Positions the patient in the supine (lysing down)
Follows the bolus (watch as food goes down)

Videofluoroscopic Swallow Study (VFSS):
Assessment of swallowing
Examines the oral cavity
Uses small amounts of liquid
Positions patient in the upright position
Does not follow the bolus

ALTERNATIVE TYPES OF FEEDING:

Nasogastric Tube - For short periods and for supplementation. This may not be a safe method for a child with a compromised swallow because of increased secretions produced by nasogastric tube, which can increase risk of aspiration.

Gastrostomy Tube or Button - Gastrostomy tube or button is a preferred choice for long-term supplementation if stomach function is intact.

Jejunostomy Tube - Preferred choice if severe gastroesophageal reflux is present.

Effects of Tube Feeding
1. Hypersensitivity of the oral cavity - as the mouth is not being stimulated, child gets sensitive to anything that is put in the mouth. Desensitizing the oral cavity is important. Always encouraging textures, tastes and sensitizing procedures even if your child is not feeding orally. (Seek the expertise of an occupational or speech therapist.)
2. After tube feeding is started even for a short period of time it is difficult to get the child back on to oral feeds.
3. Lumps and different textures may remain a problem into later childhood.
4. The tube may not work, dislodge, and need replacing. Granulation tissue (healthy scar tissue) can collect around the tube and so can infection.
5. Parents like the button as it gives their child better mobility and freedom to move.

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THE CHARGE ADOLESCENT - PHYSICIAN INFORMATION

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The adolescent period is a stage of change and offers its own challenges for all families but especially to those with CHARGE teens.

Behavioral problems can be a big issue. These may be outbursts, tantrums, self-abuse, and defiance.[1] For a non-CHARGE adolescent unruly behavior is more likely due to psychological concerns than from medical ones. With the CHARGE adolescent a medical cause needs to be considered first especially if these outbursts occur without any provocation or warning and are waking the adolescent from sleep[3].

There are certain co-morbid psychological conditions that can occur with CHARGE. These may be detected before adolescents and these include:

- Attention deficit disorder (ADHD)
- Pervasive developmental disorder (PDD)
- Obsessive compulsive disorder (OCD)
- Anxiety
- Depression
- Learning difficulties

If it is suspected that the CHARGE adolescent has one of these co-morbid conditions secondary to the CHARGE diagnosis, it is important to have a full psychological evaluation including a detailed home and school profile. The psychologist should have understanding of adolescents with sensory deficits. It is important to take care of the above labeled diagnosis as it is easy to overlook sensory deficits i.e. hearing and visual loss.

Drugs used to treat these co-morbid conditions may be required in smaller doses than for non-CHARGE adolescents. Before starting medication, it is best to treat the adolescent not the label. Any of the listed diagnoses often gives rise to adverse behaviors but so do frustrations in not being able to communicate or frustration in this non-CHARGE world. It is important to observe when these types of behavior are occurring and to try behavior modification techniques along with a very structured day.[4] Planning a daily routine and preparing the adolescent for any change may be a good strategy. Positive rewards for good behavior and a routine are a very good start to a behavioral modification program. The adolescent with CHARGE responds like many other non-CHARGE adolescents and would benefit from having a set of rules about behavior that is and is not acceptable,[1] e.g. The parent may tell their teen they can touch a persons hand, shoulder and arm, but that they are not allowed to touch peoples breasts.[1]

Observations about behavior in CHARGE adolescents directly from a handout from Veronika Bernstein[1] are as follows:

✓ *Denotes behaviors which can occur in any adolescent.*

- ✓ mood disturbances
- ✓ marked irritability
- ✓ emotional lability
- ✓ anxious
- ✓ depressed

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- ✓ insomnia
- ✓ excessive sleepiness
- ✓ eating problems

diminished taste
changes in cognition

- ✓ changes in concentration

deterioration in vision and hearing related to not paying attention
slowing of mental processing

- ✓ diminished initiative

impaired memory

- ✓ increased in impulsivity

repeating same sign or phrase over and over
obsessive-compulsive rituals
tics starting with facial grimaces to major muscle groups

- ✓ increased aggression

increase in self-injurious behaviors
increase in sensation seeking

- ✓ novel experiences
- ✓ intense experiences
- ✓ increase in risk taking behaviors
- ✓ violation of well learned rules

- ✓ increase in thoughts of invincibility and telepathic powers

interest in sexually related behaviors:
increase in masturbation
increase in staring at others (inappropriate)
increase in touching others (inappropriate)

- ✓ hate to be corrected
- ✓ hate to be told what to do
- ✓ hate to be bored
- ✓ hate to be bothered

The above could describe any typical adolescent - with or without CHARGE.

If the adolescent is experiencing regression in behavior or cognition (mental processess - memory), it is important to rule out an underlying medical deterioration i.e. hypothyroidism, heart disease, regression in hearing and vision.

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THE CHARGE ADOLESCENT - PARENT INFORMATION

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Two key things to remember about your adolescent:

Their success and performance will vary from day to day. Just because they accomplished something yesterday, don't look down upon them because they can't do it today.[1]

There is no miracle strategy for dealing with your adolescent. Just keep trying new things until you find something that works with your teen.

MEDICAL ISSUES:

An important aspect of your child's adolescents is the onset of puberty. Both males and females with CHARGE may require hormone replacement therapy. If a deficiency in sexual hormones is suspected, an appointment with an endocrinologist is suggested.[5] Many families forget the "sex hormone" issues and are either never referred to an endocrinologist or go too late.

In males, androgen replacement therapy (male sex hormones) may cause significant growth in the penis. The psychological boost derived from this treatment is very significant and sufficient reason for considering this therapy. Males also tend to have poor growth of facial hair, which may be alleviated by this therapy[5].

Females without hormone replacement therapy tend to enter puberty late and have irregular periods. They may also have no breast development without estrogen replacement therapy.[2]

As more CHARGE children enter adolescents there is an increased awareness of problems and how to approach these. Skeletal curvature (scoliosis) is of concern and was brought to our attention when conducting a survey at the 1997 Family College Association Conference.[4] If there is concern, ask to see an orthopedic surgeon. This scoliosis may increase during the growth spurt of the adolescent (13-20 years). The growth spurts in CHARGE adolescents occur later than the documented norms for growth. This puberty pattern reflects the potential for later growth and for a normal adult stature.

As many CHARGE children obtain milestones late (walking, running, communicating), it is not unreasonable to suspect that many of them crave for more learning opportunities later in life. These need to be respected and as some of our adolescent case studies show it is not reasonable to expect the adolescent to be happy doing "basket weaving" or other menial tasks.

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LIFE EXPECTANCY IN CHARGE

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Children with CHARGE have been shown to have a 70% survival rate to five years of age (from actuarial survival analysis).[1,2] The death rate is the highest in the first year of life.

The highest mortality is seen with a combination of CHARGE features of bilateral posterior choral atresia with either congenital heart defects or tracheo-esophageal.[1] If all three of the above are present, they offer the greatest risk of mortality or morbidity to the infant.

Patients with CHARGE have a high postoperative mortality.[3] The reasons for this are postulated as hidden structural abnormalities of the larynx and/or pharynx with associated motor incoordination. This results in difficulty in intubation and problems after extubation. It is therefore important that the surgeons and anesthetists be aware of how complex these children are. When contemplating surgery in these children an experienced pediatric anethesiologist should be involved, even if the surgery is rated as a minor procedure. It is advised that, if possible, a number of procedures be conducted under one anesthetic, thus limiting the number of anesthetics. A recent CHARGE girl at our hospital (IWK Grace Health Center, Halifax, Nova Scotia) undertook seven operations at the same time under one anaesthetic so it can be done!

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CHARGE SYNDROME: GLOSSARY
Compiled by Meg Hefner, M.S.

A

Accutane: (isotretinoin, retinoic acid): prescription medication used to treat severe cystic acne. It is a synthetic derivative of Vitamin A. When taken during pregnancy, it can cause very serious birth defects, including hydrocephalus, microcephaly (very small head), mental retardation, small and malformed ears and other facial abnormalities, and heart defects. Although the ear malformations can be similar to those seen in CHARGE, the face is distinct.

amblyopia: "lazy eye" - poor vision in one eye without detectable cause. Often treated by patching the stronger eye.

anesthesia: 1. Loss of sensation resulting from pharmacologic depression of nerve function or from neurological dysfunction. 2. Sedation used during surgery.

anophthalmia: absence of the eye or eyes. Anophthalmia can be considered the most severe form of a coloboma.

aorta: the large artery arising from the base of the left ventricle of the heart. The aortic arch is formed by the ascending aorta and the descending aorta.

aortic arch: the curved portion between the ascending and descending parts of the aorta. Aortic arch anomalies are common in CHARGE

apnea: a potentially life-threatening condition inn which breathing stops abnormally, usually during sleep. CPR is sometimes needed to start the breathing again.

arhinencephaly: a term used to refer to an absence of some or all areas of the forebrain, or certain areas of the anterior front) portions of the brain, particularly the olfactory bulbs and nerves. The defect may occur relatively late in fetal differentiation, so that only a small area of the brain is involved. One result can be the lack of a sense of smell.

aspiration: the inspiratory sucking into the airways of fluid or foreign body, as in vomitus. Children with CHARGE are at risk for aspiration of food or liquid which is not adequately swallowed or which is refluxed up from the stomach.

aspiration pneumonia: a lung inflammation caused by inhaling a foreign body, such as food, into the lungs.

association: a connection of persons, things or ideas by some common factor. In genetics: the occurrence together in a population, more often than can be readily explained by chance, of two or more traits of which at least one is known to be genetic. For example VACTERAL association.

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atresia: a general term for closure of a structure that should be open, such as a canal, passage or tube. See choanal atresia, esophageal atresia, and ear anomalies.

atrial septic defect (ASD): hole between the two upper chambers (atria) of the heart. Surgery is usually required to close an ASD.

audiogram: the graphic record drawn from the results of hearing tests with audiometer. The audiogram charts the threshold of hearing at various frequencies (pitch) against sound intensity in decibels (loudness). Pure tone audiogram: a chart of the threshold for hearing acuity at various frequencies usually expressed in decibels above normal threshold and usually covering frequencies from 128 to 8000 Hz. Speech audiogram: the record of thresholds for spondaic word lists and scores for phonetically balanced word lists. See Ears and Hearing Section.

autistic disorder: impaired development of social interaction and communication along with repetitive and stereotyped patterns of behavior, interests, and activities, with an onset prior to age three.

austitic behaviors may include withdrawal from social contact, avoidance of eye contact, and failure to develop friendships; delay or total lack of communication, or communication is abnormal; self-stimulation, preoccupation with objects or parts of objects, obsession with certain routines or rituals. Austistic-like behaviors are observed in some children with CHARGE. It is not clear whether to attribute them to central nervous system structural abnormalities, to dual sensory impairment, or whether a separate diagnosis of autism is appropriate.

B

BAER: abbreviation for "brainstem auditory evoked response." Measurement of alteration in the electrical activity of the auditory system of the brain brought about by presenting sounds through earphones. This can be done with the child sedated and without the cooperation of the child. Also called ABR.

bilateral: two-sided, or affecting both sides.

C

cartilage: connective tissues found primarily in joints, the walls of the thorax, and tubular structures such as the larynx, air passages, and ears; comprises most of the skeleton in early fetal life, but is slowly replaced by bone. Cartilage is often weak in children with CHARGE, resulting in floppy ears, laryngomalacia and other complications.

Cardiologist: physician who specializes in the heart.

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CATCH-22: another name for VCF (velocardiofacial syndrome), which has some features which are similar to features seen in CHARGE, but is due to a microdeletion of chromosome 22.

central auditory processing: a central hearing loss that may prevent sound from being interpreted meaningfully by the brain. Sound may be picked up by the ears and signals transmitted to the brain, but the brain has difficulty making sense of the input.

central nervous system (CNS) abnormalities: CNS abnormalities seen in children with CHARGE Syndrome include: structural brain abnormalities (diagnosed by CT scan or MRI), microcephaly, seizures, apnea, and central processing problems (including a central hearing loss). See also: cranial nerve abnormalities and Brain Section.

cerebellum the large back portion of the brain. It consists of the two lateral hemispheres united by a narrow middle portion, the vermis.

cerebrum originally referred to the largest portion of the brain, including practically all parts within the skull except the medulla, pons, and cerebellum; it now usually refers only to the parts derived from the telencephalon and includes mainly the cerebral hemispheres (cerebral cortex and basal ganglia.

cerumen: ear wax

CHARGE facial features: Children with CHARGE Syndrome resemble their parents, but they may also have some of the following features that make them look similar to other children with CHARGE Syndrome: square shape of the face and head, flat cheekbones, facial asymmetry (with or without facial palsy), wide nose with high bridge, and unusual ears (see Ear anomalies). The facial features do not cause any health problems, but can be very helpful in making the diagnosis of CHARGE Syndrome. See Diagnosis Section.

choroid (eye): the portion of the eye lying between the retina and the sclera.

chromosomes: microscopic structures found in the nucleus of cells which contain the genetic information (DNA). Possible chromosome tests include routine (g-banded, including most prenatal chromosome testing), fragile X (to diagnose a particular cause of mental retardation), and FISH (to look for specific microdeletions or other abnormalities). No specific chromosome abnormalities have yet been shown to cause CHARGE. In VCF, there is a small deletion (microdeletion) of chromosome 22.

choanal atresia or stenosis: The choanae are the passages from the back of the nose to the throat which allow breathing through the nose. Choanal atresia is a birth defect in which this passage is completely blocked; stenosis refers to a narrowed passage. The atresia or stenosis can be on one or both sides. If a baby is born with bilateral choanal atresia, immediate surgery is needed to permit the baby to breathe through the nose. Choanal stenosis can lead to problems with breathing or increased nasal stuffiness. See Choanal atresia Section.

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cleft lip and/or cleft palate: Many children with CHARGE Syndrome have a cleft lip (hare lip) and/or cleft palate (an opening in the roof of the mouth). Children with a cleft palate may have problems with ear infections and speech even after surgical correction. Submucous cleft palate may be difficult to diagnose in children with CHARGE. See Cleft lip and palate Section.

cochlea: part of the inner ear, responsible for transmitting sound to the auditory nerve.

cochlear implant: a device that changes sound into electrical signals. These signals are then sent through the skin to an electrode array surgically implanted within the cochlea. Some individuals with severe to profound sensorineural hearing loss can get some hearing through a cochlear implant. Experience with cochlear implants in children with CHARGE is limited.

coloboma: a cleft or keyhole-shaped defect of the eyeball. Colobomas can occur anywhere in the eye (any combination of iris, retina, or disc) and can affect one or both eyes. A coloboma of the iris (colored part of the eye) will result in a keyhole-shaped pupil, but probably will not affect vision. A coloboma of the retina or disc in the back of the eye can only be detected by an eye exam done by an experienced ophthalmologist. Retinal or disc colobomas can cause significant vision loss, both by restriction of the visual field (large blind spots across the top of normal vision) and by decreased acuity (blurred vision). See Eye Section.

columella: the part of the nose between the nostrils. It is sometimes prominent in children with CHARGE. See CHARGE face photos in Diagnosis Section.

conductive hearing loss: Hearing loss due to abnormalities of the middle ear bones (ossicles) and/or to fluid accumulation in the middle ear. If the loss is due to fluid accumulation, it may get better with use of PE tubes and/or decongestants. If the loss is due to malformed ossicles, surgery might be considered, but this is controversial.

conotruncal anomalies of the heart: the most common class of heart defects in CHARGE, including, double outlet right ventricle, tetralogy of Fallot, interrupted aortic arch, and VSD.

corpus callosum: the plate of nerve fibers which connect the two cortical hemispheres of the brain. Some individuals with CHARGE have absent or hypoplastic (underdeveloped) corpus callosum.

cranial nerves: those nerves that emerge from, or enter, the cranium or skull, in contrast to the spinal nerves, which emerge from the spine or vertebral column. The twelve paired (one on each side) cranial nerves are the I - olfactory, II- optic, III - oculomotor, IV - trochlear, V - trigeminal, VI - abducent, VII - facial, VII - vestibulocochlear, IX - glossopharyngeal, X - vagal, XI - accessory, and XII - hypoglossal. Cranial nerve abnormalities are very common in CHARGE.

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cryptorchidism: undescended testicles. If the testes do not descend into the scrotum, they can be lowered surgically. (Existing definition)

CT scan: computerized tomography. A special X-ray of the head used to look at the structure of the brain.

D

Dandy-Walker malformation or cyst: developmental anomaly of the fourth ventricle of the brain. It can result in cerebellar hypoplasia, hydrocephalus, and posterior fossa cyst formation.

developmental delay: most children with CHARGE Syndrome will have delayed or retarded development. Vision loss causes delays in motor development. hypotonia (low muscle tone) and balance problems also delay motor development. Hearing loss can cause delays in speech and language. As developmental depends on the combination of intelligence, hearing, and the ability to see, early developmental delay does not always mean mental retardation (see mental retardation). Many children with CHARGE have normal intelligence.

DiGeorge sequence: congenital absence of the thymus and parathyroid glands. This leads to increased infections and delayed development. Common in VCF syndrome, rare in CHARGE.

dysfunction: difficult or abnormal function

E

ear anomalies: Children with CHARGE Syndrome often have misshapen ears. Often the ears have characteristics which are unique to CHARGE. Typical "CHARGE ears" are small and wide, with little or no ear lobe. Often the outer fold of the ear (helix) is missing or may appear clipped-off. The child's two ears often look different. The unusually shaped ears do not cause hearing loss unless there is stenosis (narrowing) or atresia (collapse) of the ear canal, which is rare in CHARGE Syndrome. If a hearing aid is required, the ear anomalies may make it difficult to fit the ear mold properly. The ear anomalies can be very helpful in making the diagnosis of CHARGE Syndrome because they look different from ear anomalies seen in any other syndrome. See EARS and Heating and Diagnosis Sections.

echocardiogram the ultrasonic record obtained by echocardiography. Sound waves are used to get an image of the heart.

EEG (electroencephalogram): brain wave test used to look for seizure activity.

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Endocrinologist: physician who specializes in treatment of hormone abnormalities.

esophageal atresia: the esophagus (food pipe) ends in a pouch instead of connecting to the stomach. Babies with this abnormality spit up all their food until it is surgically corrected. Even after surgery, feeding may be difficult for some time due to other swallowing problems which are seen in CHARGE Syndrome. See TEF/EA Section.

eustachian tube: the tube leading from the tympanic cavity (behind the eardrum) to the nasopharynx (back of the throat). It enables equalization of pressure within the tympanic cavity with ambient air pressure, referred to commonly as "popping of the ears." Blocked eustatian tubes predispose to ear infections.

evoked potentials auditory screening for newborns using auditory evoked potential measures, including auditory brainstem response (ABR, BAER)

exotropia: outward turning of the eyes.

F

facial palsy or paralysis: The facial nerve (cranial nerve VII, which controls facial muscles) does not work in many children with CHARGE. This can cause a lopsided smile and trouble blinking the eye on the affected side. The affected eye may not produce tears very well. It may be unilateral or bilateral. If bilateral, it leads to a very blank expression.

FISH Fluorescent in-situ hybridization: a specialized chromosome test using fluorescent dyes to identify very small deletions or other abnormalities of chromosomes. Many children with CHARGE will have chromosome tests done including FISH for deletion of chromosome 22 seen in VCF.

FM Trainer: FM (frequency-modulated) signal transmission, represents the most successful and largest market of assistive listening devices for children. FM systems have an advantage of an improved signal-to-noise ratio over hearing aids.

fundoplication: An operation in which the opening from the esophagus to the stomach is tightened to prevent gastroesophageal reflux.

G

G-tube: gastrostomy tube.

Gastroenterologist: physician who specializes in the esophagus, liver, spleen, bowel and pancreas.

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gastrostomy: an operation in which an artificial opening is made into the stomach through the wall of the abdomen. The child is then fed through the gastrostomy tube instead of orally.

gastrostomy tube: a tube which is surgically placed in the stomach through the abdominal wall for feeding. This is done when the child cannot eat by mouth because of a severe cleft palate, tracheo-esophageal (T-E) fistula, or swallowing problems. Often, a gastrostomy button or Mic-key is used.

GE reflux: gastroesophageal reflux, the backward flow of food from the stomach into the esophagus, possibly into the pharynx where they can be aspirated between the vocal cords and down into the trachea; symptoms of burning pain and acid taste result; pulmonary complications of aspiration are dependent upon the amount, content, and acidity of the aspirate. This is very common in CHARGE.

growth deficiency: small stature or size. In CHARGE, this can be due to multiple medical problems, growth hormone deficiency, or some other cause

growth hormone deficiency: somatotropin, a protein hormone of the anterior lobe of the pituitary, produced by the acidophil cells, that promotes body growth, fat mobilization, and the inhibition of glucose utilization; a deficiency of, is associated with a number of types of dwarfism. Rare, but not unheard of in CHARGE.

H

hearing loss: About 85% of children with CHARGE Syndrome have some hearing loss. The loss can be conductive, sensorineural (nerve), or mixed (both) and can range from a mild hearing loss to profound deafness. The loss may be progressive. A CHARGE Syndrome hearing loss is typically a mixed loss with a large conductive component in the low frequencies and a sensorineural loss or mixed loss for high frequency sounds. The losses can be severe and very difficult to measure completely and accurately, especially in infants and young children. As a result, repeated testing may be required before a satisfactory set of results is obtained. See Ears and Hearing Section.

heart disease, congenital (CHD): About 2/3 of children with CHARGE are born with some kind of heart defect. Some have only a murmur, while others may have a life-threatening heart defect which requires surgery. Heart abnormalities described in children with CHARGE include tetralogy of Fallot, ASD, VSD, PDA, aortic arch anomalies, double outlet right ventricle, pulmonic stenosis, and others. All children in whom a diagnosis of CHARGE is suspected should have a cardiac evaluation.

hockey-stick crease: crease on the palm of the hand which bends up to form a deep crease between the index and middle fingers. Although this has no medical significance, it is very common in children with CHARGE. See photo in Diagnosis Section.

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holoprosencephaly: failure of the forebrain or prosencephalon to divide into hemispheres or lobes; cyclopia occurs in the severest form. It is often accompanied by a deficit in midline facial development. Often diagnosed by MRI or CT. Rare in CHARGE.

horseshoe kidney: union of the lower or occasionally the upper extremities of the two kidneys by a band of tissue extending across the vertebral column, resulting in a single horseshoe-shaped kidney. See Urinary/Renal Scetion.

hydrocephalus: a condition marked by an excessive accumulation of fluid in the brain, resulting in dilation of the ventricles of the brain and raised intracranial pressure; may also result in enlargement of the cranium and atrophy of the brain. Treated by surgical placement of a shunt. Rare in CHARGE.

hydronephrosis ("water on the kidneys"): dilation of the pelvis and calices of one or both kidneys resulting from obstruction or backflow of urine. If not corrected, this can cause kidney damage or failure. Common in CHARGE.

hypogonadism: underdeveloped genital system. In boys, there may be a small penis or cryptorchidism (undescended testicles). In girls, the labia (external skin folds) may be small or absent. In children of both sexes, hypogonadism may prevent puberty unless hormones are given.

hypoplasia: underdevelopment of a tissue or organ.

hypospadias: a genital problem in males. The urethral opening is not at the end of the penis and needs to be corrected surgically.

hypotonia: low muscle tone. This can lead to a "floppy" baby. Many children with CHARGE have hypotonia, especially of the trunk (upper body), contributing to delay of some motor milestones such as sitting and walking and predisposing the children to scoliosis.

I

IEP Individualized Education Plan

imperforate anus: the anus is closed over and needs to be opened surgically. Sometimes only a thin membrane needs to be opened. More often the blind end of the large bowel needs to be connected to the skin on the belly as a colostomy. Later the end of the bowel is put down through an artificial opening created where the anus ought to be. Not the same as choanal atresia. This is not usually seen in CHARGE.

incontinence: inability to prevent the discharge of any of the excretions, especially of urine or feces. Includes bedwetting.

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infections: Children with CHARGE Syndrome tend to be very sickly, especially in the early years. The most common illnesses are recurrent ear infections and pneumonia. The pneumonias tend to become less frequent after two to three years of age. The ear infections and ear drainage may last well into the teens.

inguinal hernia: also known as a "rupture". This is a lump noted in the groin and is actually a small loop of intestine sticking out a small hole connecting the inside of the abdomen to the groin. If the intestine gets stuck there, gangrene can occur, so preventive surgery is necessary.

Iris: the colored part of the eye, with the pupil in the center. A coloboma of the iris results in a keyhole-shaped pupil. Common in CHARGE.

IUGR: Intrauterine growth retardation. Slower than expected growth of a baby before birth. At birth, it will show up as low birth weight.

K

kidney abnormalities: 40% of children with CHARGE have kidney abnormalities. Kidney abnormalities seen in CHARGE include hydronephrosis, small or absent kidney, posterior urethral valves, and kidney reflux. See Kidney Section.

kidney reflux: back flow of urine into the kidney. This can result in eventual damage to the kidney. IVP is often needed to confirm reflux in the kidneys.

L

labia: female genital folds. May be smaller than normal in girls with CHARGE.

laryngomalacia or chondromalacia of larynx: the presence of soft laryngeal cartilage, most often seen in the epiglottis of young children. Very common in CHARGE, and contributes to surgical risks of anesthesia, breathing problems and swallowing problems.

larynx: the organ of voice production; the part of the respiratory tract between the pharynx and the trachea; it consists of a framework of cartilages and elastic membranes housing the vocal folds and the muscles which control the position and tension of these elements. May be weak in CHARGE.

limb defects: abnormalities of arms, legs, hands, or feet. Occasional limb defects seen in CHARGE include thumb and forearm abnormalities. See Skeletal Section.

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M

macula: the center of the retina of the eye. The macula is responsible for seeing details. Coloboma of the retina can result in legal blindness.

Medical Geneticist: physician with medical training (most often in pediatrics or OB/Gyn) with subspecialty training in Medical Genetics or (more recently) physician with residency training in medical genetics and certification by the American Board of Medical Genetics of the AMA.

mental retardation: The intelligence in children with CHARGE Syndrome ranges from normal to severe mental retardation. Most children with CHARGE have some intellectual limitations. Intelligence is very hard to estimate and in fact is often underestimated when these children are young. This is because vision and hearing problems can delay speech and development and because so many of these children spend a large portion of their early years in the hospital. Nevertheless, it is important to have your child evaluated early and often to help set up the most appropriate educational program possible. Evaluations should be done by specialists with experience testing children with sensory deficits (hearing and vision loss).

microcephaly: unusually small head

micrognathia: small jaw or chin

micropenis: abnormally small penis. See Genital Section.

microphthalmia/microphthalmos: abnormal smallness of the eye. A severe coloboma can result in microphthalmia or anophthalmia.

midface: the middle of the face, especially the cheekbone area. May be flattened or small in CHARGE.

Mondini defect: an abnormal opening from the semicircular canal into the middle ear. This is a potentially treatable (with surgery) cause of balance problems.

MRI: magnetic resonance imaging; a diagnostic form of imaging (of the brain or other body parts) Unlike conventional radiography or CT, MRI does not expose patients to ionizing radiation. In addition, it can provide superior 3-D images of the body's interior, delineating muscle, bone, blood vessel, nerve, organ, and tumor tissue. Often used to describe the exact choanal atresia and/or inner ear abnormalities in CHARGE.

N

nares: nostrils. May be small or appear pinched in CHARGE.

Neurologist: physician who specializes in the nervous system.

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NG tube: nasogastric tube. Feeding tube which is put through the nose, down the throat into the stomach for feeding.

nystagmus: involuntary movements of the eyeball, most often side to side or in circles.

O

olfactory nerve: cranial nerve I, responsible for smell. Often absent or abnormal in children with CHARGE.

omphalocele: failure of the abdominal wall to close properly around the umbilical cord during fetal development. Often some of the intestines are outside the body. This can usually be corrected surgically. An umbilical hernia is a very mild omphalocele.

Ophthalmologist: MD who specializes in the eyes. Children with CHARGE should be evaluated by a Pediatric Ophthalmologist.

optic nerve: cranial nerve VII. Abnormalities of cranial nerve VII result in facial palsy.

optometrist: technician who specializes in detecting problems of visual acuity.

ossicles: tiny bones in the middle ear: anvil, stapes and hammer.

ossicular malformation: malformation of the small bones of the middle ear (hammer, anvil and stapes). Ossicular malformations are very common in CHARGE and result in conductive hearing loss.

OT: occupational therapist. A specialist in the development of fine motor, social, and adaptive skills.

otitis media: ear infections. These can occur when fluid accumulates in the middle ears, behind the ear drums. If the fluid is infected, the child will complain of pain and there will be hearing loss. If the fluid is not infected, the child usually will have no pain, but may still have significant hearing loss. In CHARGE Syndrome, otitis media often lasts into the teenage years and requires constant medical care (see PE tubes).

Otolaryngologist: specialist in hearing, either MD (ENT specialist) or PhD (Audiologist)

P

PAX2: group of genes responsible for orchestrating development of eyes, ears, and kidney. No PAX2 gene abnormalities have yet been detected in children with CHARGE.

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palmar crease: referring to the palm of the hand. Children with CHARGE often have an unusual "hockey-stick" palmar crease. See photos in Diagnosis Section.

parathyroid gland: adjacent to the thyroid gland; one of two small paired endocrine glands. They secrete parathyroid hormone that regulates the metabolism of calcium and phosphorus.

PE tubes: tiny polyethylene (plastic) tubes which can be surgically placed in the ear drum to drain the excess fluid from behind the drum. This will help prevent hearing loss caused by recurrent otitis media. Often several sets are needed over many years in children with CHARGE.

perseverative behavior: repetitive, repeated behavior, often seen in children with sensory deficits.

photophobia: intolerance to light, especially bright lights

pinna: external ear. Often very unusual in CHARGE.

pituitary abnormalities: The pituitary is a gland at the base of the brain which produces several important hormones which help control growth, thyroid and sex gland function, and steroid production. The pituitary gland does not function properly in some children with CHARGE. This can result in deficiencies in growth hormone and in the sex hormones which cause puberty. If left untreated, these children will be short and will not develop secondary sex characteristics. Hormone therapy is available for these problems.

polyhydramnios: excess amount of amniotic fluid. In pregnancies where the baby has CHARGE, polyhydramnios can related to choanal atresia and/or swallowing abnormalities.

postnatal: occurring after birth

posterior urethral valves: small pieces of tissue which prevent urine from flowing out of the bladder through the urethra to the outside. This can cause back-up of urine and damage to the kidneys. Surgery is usually necessary.

prenatal: preceding birth; antenatal

proprioception: A sense or perception, usually at a subconscious level, of the movements and position of the body and especially its limbs, independent of vision; this sense is gained primarily from input from muscles and the vestibular (balance) apparatus.

ptosis: droopy eyelids, caused by a facial nerve problem in CHARGE syndrome.

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R

renal: nephric; relating to the kidney.

retarded development: see Developmental delay and Mental retardation.

retina: the part of the eye which receives light and transmits electrical signals to the brain, resulting in vision. Colobomas of the retina will result in blind spots and/or visual acuity (sharpness) abnormalities.

retinal detachment: separation of the retina from the choroid. This can result in blindness.

retinoic acid Vitamin A acid; used topically in the treatment of acne. See Accutane.

S

sclera: whites of the eye

sedation: medication given to calm and/or quiet a child to make testing easier and/or more accurate. Children with CHARGE can have unusual (more or less extreme) reactions to sedation.

semicircular canals: part of the inner ear involved in balance

sensorineural hearing loss (nerve deafness): hearing impairment due to disorders of the cochlear division of cranial nerve VIII (auditory nerve), the cochlea, or the mitochondrial nerve tracts, as opposed to conductive hearing loss. Very common in CHARGE.

sensory deficit: vision loss and/or hearing loss.

sonogram: ultrasound

short stature: can be caused by a lack of growth hormone (see pituitary abnormalities). Some children with CHARGE Syndrome have normal levels of growth hormone but still have short stature from an unknown cause.

stapes: the smallest of the three auditory bones in the middle ear.

stapedius tendon: stapedius muscle; dampens vibration of the stapes by drawing head of stapes backward as a result of a protective reflex stimulated by loud noise. The stapedius tendon is often abnormal in CHARGE.

strabismus: crossed eyes

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stridor: a high-pitched, noisy respiration, like the blowing of the wind; a sign of respiratory obstruction, especially in the trachea or larynx.

swallowing difficulties: Many children with CHARGE Syndrome have trouble coordinating the muscles used for normal sucking and swallowing, even in the absence of other problems such as esophageal atresia or T-E fistula. This may be due to abnormalities in cranial nerves IX and X. The incoordination can lead to gagging, apnea (breathing stops), and pneumonia (food inhaled into the lungs, causing infection). In some cases, tube feeding (gastrostomy) is used until the child is able to learn to swallow. Children with CHARGE may not learn to swallow effectively until they are 5 or 6 years old. See Feeding Section.

syndrome: the aggregate of signs and symptoms associated with any morbid process, and constituting together the picture of the disease. As in CHARGE syndrome.

T

teratogen: a drug or other agent that causes abnormal fetal development, for example, Accutane. There are no teratogens known to cause CHARGE.

thymus: a gland in the lower part of the neck, that is necessary in early life for the normal development of immunological functions. It reaches its greatest relative weight shortly after birth and its greatest absolute weight at puberty; it then begins to involute, and much of the lymphoid tissue is replaced by fat. It is abnormal in children with DiGeorge sequence.

ToF: Tetralogy of Fallot. Complex heart defect often seen in CHARGE.

tracheo-esophageal (T-E) fistula: an abnormal connection between the trachea (wind pipe) and esophagus (food pipe). If it is not corrected surgically, food will get into the lungs and cause choking and/or pneumonia.

tracheomalacia: weakness of elastic and connective tissue of the trachea. Common in CHARGE. Can cause complications with anesthesia.

tracheostomy/tracheotomy: the operation of opening into the trachea to permit breathing directly into the throat, usually intended to be temporary. Often necessary in CHARGE.

U

ultrasound: imaging procedure which uses high-frequency sound waves to get an image of a fetus during pregnancy. Echocardiography also uses ultrasound to image the heart.

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umbilical hernia: see omphalocele.

unilateral: affecting one side only.

URI: upper respiratory infection

urinary tract malformations: The urinary tract malformations seen in CHARGE Syndrome include posterior urethral valves, abnormal kidney shape or location, and backup of urine from the bladder into the kidney (reflux).

UTI: urinary tract infection

VACTERAL/VATER association Acronym for Vertebral defects, Anal atresia, Cardiac defects, TracheoEsophageal fistula with esophageal atresia and Renal anomalies and Limb defects. Many features overlap with those of CHARGE, but can be distinguished by a Medical Geneticist.

VCF: velocardiofacial syndrome; Shprintzen syndrome; CATCH-22. Syndrome consisting of cleft palate, heart defects, learning disabilities, and distinct physical features are all present. The overwhelming majority of children with VCF have a microdeletion of chromosome 22 detectable by FISH. Although many features overlap, it is distinct from CHARGE, and the shapes of the ears, face, and hands are distinct from CHARGE. To date, no one with a definite diagnosis of CHARGE has been positive for the chromosome 22 deletion associated with VCF.

velopharyngeal: pertaining to the soft palate (velum palatinum) and the posterior nasopharyngeal wall.

ventricles (head, heart): A normal cavity or space. In the heart, the spaces through which blood is pumped (left and right ventricle). In the brain, fluid-filled spaces (lateral, third, and fourth ventricles) (see hydrocephalus).

vestibular: relating to a vestibule, especially the vestibule of the ear. The vestibule of the ear is responsible for one component of balance. Vestibular abnormalities are common in CHARGE and contribute to delay in motor milestones such as sitting and walking.

VER: visual evoked responses;

videofluoroscopy: examination x-ray, using the fluoroscope, using an image intensifier and television camera for image detection and a video monitor for display to get live action pictures of , e.g. swallowing.

visual field: the portion of side view, and top and bottom view, in which a child can see or has functional vision. Colobomas cause visual field defects (blind spots).

webbed neck: wide or broad neck, often somewhat short. Many children with CHARGE have a webbed neck or just a short neck.

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CHARGE Syndrome Resources

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CHARGE Syndrome Foundation, Inc.
2004 Parkade Blvd.
Columbia, MO 65202-3121
phone: (800)442-7604 Parent line (USA callers only)
(573)499-4694 all others
FAX: (573)499-4694
e-mail: marion@chargesyndrome.org
http://www.chargesyndrome.org, http://www.kumc.edu/gec./support/charge.html

INTERNATIONAL CHARGE ORGANIZATIONS

CHARGE Family Support Group
115 Boundary Road
Colliers Wood,
London SW192DE
United Kingdom
phone: (01)81 540-2142

The Australasian CHARGE Association
P.O. Box 91
Glenfield, N.S.W. 2167
Australia
phone: (02) 9829-4801
e-mail: austcharge@hotmail.com

Queensland CHARGE Association
16 Ronmack Street
Chermside QLD 4032
Australia
phone: (07)3359-6297
e-mail: charge_qld@hotmail.com
http://www.users.bigpond.com/arrone/

** CHARGE WEB PAGE and LISTSERVE**

CHARGE Syndrome Foundation, Inc.
http://www.chargesyndrome.org

Minnow's CHARGE WebPage:
http://www.geocities.com/Heartland/1220/
charge@saber.net

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"DEAF AND DEAFBLIND RESOURCES"

A.G. Bell Association for the Deaf
3417 Volta Place, N.W.
Washington, DC 20007-2778
phone: (202) 337-5220 (v, TTY)
http://www.agbell.org/

American Society for Deaf Children
PO Box 1510
Olney, MD 20830
Attn: Mr. Ken Silverstein
1-800-942-ASDC Parent line
http:www.deafchildren.org/
ASDC1@aol.com

DB-LINK, The National Information Clearinghouse on Children who are Deaf-Blind.
(Also lists 307.11 State grantees Deaf-Blind projects)
Teaching Research
345 N. Monmouth Ave.
Monmouth, OR 97361
phone: (800)438-9376 (voice), (800)854-7013 (TTY), FAX: (503)838-8150
e-mail: dblink@tr.wou.edu
http://www.tr.wou.edu/dblink/index.htm

Deafblindness Web Resource
http://www.deafblind.co.uk
Includes terminology, communication, journals, periodicals

The Family Centre
86 Cleveland Road
Ealing, London W13 OHE
United Kingdom
phone: (01)81 991-0513, FAX: (01)81 810-5298

The Foundation for Fighting Blindness
Executive Plaza 1, Suite 800
11350 McCormick Road
Hunt Valley, MD 21031-1014
phone: (800)683-5555 (voice), (800)683-5551 (TTY), FAX: (410) 771-9470
http://www.blindness.org

John Tracy Clinic
806 West Adams Boulevard
Los Angeles, CA 90007
phone: (213) 748-5481 (v) , (213) 747-2924 (TTY), (800) 522-4582, FAX: (213) 749-1651
http://www.johntracyclinic.org

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Helen Keller National Center for Deaf-Blind Youths and Adults (HKNC)
111 Middle Neck Road
Sand Points, NY 11050-1299
phone: (516) 944-8900 (v), (516) 944-8637 (TTY), FAX: (516) 944-7302
e-mail: abigailp@aol.com
http://www.helenkeller.org/national/index.htm
HOUSE Ear Institute
2100 West 3rd Street
Los Angeles, CA 90057
phone: (800)287-4763 (v, TTY) California, (800)352-8888 (v, TTY) all other 50 states
http://www.hei.org/

Katlyn's HOPE
303 South Elm Street
Wellington, KS 67152
phone: (316)326-6118, (877)677-HOPE – Toll-free for parents
e-mail: khope@idir.net
http://www.idir.net/~khope/links.html

National Association for Parents of the Visually Impaired, Inc. (NAPVI)
P.O. BOX 317
Watertown, MA 02471
phone: (617) 972-7441, (800) 562-6265, FAX: (617) 972-7444
http://www.spedex.com/napvi

National Coalition on Deaf-Blindness (NCDB)
175 North Beacon Street
Watertown, MA 02472
phone: (617) 972-7347, FAX: (617) 923-8076
e-mail: davies@perkins.pvt.k12.ma.us

National Cued Speech Center
PO Box 31345
Raleigh, NC 27622-1345
phone: (919) 828-1218 (v, TTY)
e-mail: ncsa@aol.com

National Family Association for Deaf-Blind (NFADB)
111 Middle Neck Road
Sand Point, NY 11050
phone: (800) 255-0411, FAX: (516) 944-5984
e-mail: pajomac@aol.com

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National Technical Assistance Consortium for Children and Young Adults who are Deaf-Blind (NTAC)
345 N. Monmouth
Monmouth OR 97361
phone: (503) 838-8096 (v) (OR), (503) 838-9623 (TTY) (OR), FAX: (503) 838-8150 (OR), (516) 944-8900 x 273 (v) (NY), (516) 883-9059 (TTY) (NY), FAX: (516) 883-9060 (NY)
http://www.tr.wou.edu.ntac

NOR-CAL Center on Deafness
Supported Employment Program
1820 Tribute Rd, Suite A
Sacremento, CA 98515
Phone: (916) 921-1045 (v,TTY), FAX: (916) 921-1177

Partners for Progress, Pre-College National Mission Program
Gallaudet University
800 Florida Ave., NE
washington, DC 20002, FAX: (202) 651-5435
http://www.gallaudet.edu/~parweb/pfp.html

SENSE The National Deafblind and Rubella Association
11-13 Clifton Terrace
Finsbury Park, London N435R
United Kingdom
phone: (01)71 272-7774, FAX: (01)71 272-6012
http://www.vois.org.uk/sense/
e-mail: enquiries@sense.org.uk

U.S. Department of Education Office of Special Education Programs, Division of Educaton Services For Children with Deaf-Blindness Program
Switzer Bldg., Room 4613
330 C Street, SW
Washington, DC 20202-2734
phone: (202)205-8165, FAX: (202)205-8971
e-mail: charles_freeman@ed.gov

DISABILITY RESOURCES

Beach Center on Families and Disabilities
3111 Haworth Hall
University of Kansas
Lawrence, KS 66045-7516
phone. (913) 864-7600 (v), (913) 864-7600 (TTY) FAX: (913) 864-7605
email: beach@dole.lsi.ukans.edu
http://www.lsi.ukans.edu/beach/beachhp.htm

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ERIC Educational Resources Information Center
Clearinghouse on Disabilities and Gifted Children
The Council for Exceptional Children
1920 Association Drive
Reston, VA 20191
phone: (800)328-0272 (v) (703) 264-9449 (TTY)
http://www.cec.sped.org/ericec/digests.htm
e-mail: ericec@cec.sped.org

ESCO Hearing Aid Insurance
Ear Service Corporation
3650 Annapolis Lane, Suite 107
Plymouth, MN 55447
http://www.earserv.com
phone: (800) 992-3726
e-mail: info@earserv.com

MEDICAID Waivers - Katie Beckett Waivers
Health Care Financing Administration (HCFA)
Within the Department of Health and Human Services (DHHS)
Family Voices National Office
PO Box 769
Algodones, NM87001
Toll-free (888) 835-5669
Voice (505) 867-2368
FAX (505) 867-6517
http://www.familyvoices.org/~vrosales/KATIEBEC.html

NICHCY (National Information Center for Children and Youth with Disabilities)
P.O. BOX 1492
Washington, DC 20013-1492
phone: (800) 695-0285 (v, TTY), 800-695-0285 (v, TTY) FAX: (202)884-8441
http://www.nichcy.org
(Among resources are: State chapters of parent and disability groups, Protection and Advocacy Agencies per State, Parent Training and Information Projects, and IDEA 1997)

NORD (National Organization for Rare Disorders)
100 Rt. 37, P.O. BOX 8923
New Fairfield, CT 06812-8923
phone: (203) 746-6518 (v), (203) 746-6927 (TTY), (800) 999-6673 FAX: (203) 746-6841
e-mail: orphan@nord-rdb.com
http://www.nord-rdb.com/orphan

Special Olympics
1325 G. Street NW, Suite 500
Washington, DC 20005
phone: (202) 628-3630, FAX: (202) 824-0200
http://www.specialolympics.org/

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TASH Disability Advocacy Worldwide
29 W. Susquehanna Ave. Baltimore, MD 21204
phone: (410) 828-8274, FAX: (410) 828-6706
http://www.tash.org/
e-mail: nweiss@tash.org

Wide Smiles Web Page
http://www.widesmiles.org/WS-58.html

Wright's Law - The Special Ed Advocate
Peter W.D. Wright, Esq.
P.O.BOX 1008
Deltaville, VA 23043
phone: (804)257-0857
http://www.wrightslaw.com/
e-mail: webmaster@wrightslaw.com

DEVELOPMENTAL and SENSORY INTEGRATION RESOURCES

Sandra L.H. Davenport, M.D.
Sensory Genetics/Neuro-Development
5801 Southwood Drive
Bloomington, MN 55437-1739
phone: (612)831-5522 (v, TTY), FAX: (612)831-0381
e-mail: slhdaven@tc.umn.edu

The HANDLE Institute
1530 Eastlake Avenue E., Suite 100
Seattle, WA 98102
phone: (206) 860-2665, FAX: (206) 860-3505
http://www.handle.org
e-mail: support@handle.org

National Academy for Child Development
P.O. BOX 380
Huntsville, UT 84317
phone: (801) 621-8606, FAX: (801) 621-8389
http://www.nacd.org/
e-mail: nacdinfo@nacd.org

Sensory Integration International (SII)
The Ayres Clinic
1514 Cabrillo Avenue
Torrance, CA 90501
phone: (310) 320-2335, FAX: (310) 320-9982
http://home.earthlink.net/~sensoryint/
e-mail: sensoryint@earthlink.net

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SPEECH, LANGUAGE AND COMMUNICATION RESOURCES

American Speech - Language and Hearing Association
10801 Rockville Pike
Rockville, Maryland, 20852
phone: (301) 897-5700 (v), (301) 897-0157 (TTY) FAX: (301) 571-0457
http://www.asha.org/contents.htm

APRAXIA-Kids website
http://www.jump.net/~gmikel/apraxia/

Listenup Web Page Kay Powell
http://members.tripod.com/~listenup/
listenup@texas.net

The Little Room
"Space and Self" book by Lilli Nielsen
Sikon Publisher
http://members.aol.com/LSWebDesgn/LNroom.html

Mayer-Johnson (Picture Communication Symbols)
PO BOX 1579
Solana Beach, California 92075
(800) 588-4548, FAX: (858) 550-0449
http://www.mayerjohnson.com/

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CHARGE Syndrome Medical Visit

Child _____

Birth Date _____

Date of Visit:	Next Visit: Time:
Specialist:	Referral to:
Specialty:	Date/Time:
Place:	Place:
Our questions: what we would like to discuss at this visit:	
Information discussed at this visit:	

U.S. Department of Education
Office of Educational Research and Improvement (OERI)
National Library of Education (NLE)
Educational Resources Information Center (ERIC)

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